

Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140

Véronique Geoffroy, Corinne Stoetzel, Sophie Scheidecker, Elise Schaefer, Isabelle Perrault, Séverine Bär, Ariane Kröll, Marion Delbarre, Manuela Antin, Anne-Sophie Leuvrey, et al.

► To cite this version:

Véronique Geoffroy, Corinne Stoetzel, Sophie Scheidecker, Elise Schaefer, Isabelle Perrault, et al.. Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39 (7), pp.983-992. 10.1002/humu.23539. hal-02371583

HAL Id: hal-02371583 https://hal.science/hal-02371583

Submitted on 19 Nov 2019

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers. L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



Whole Genome Sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140

Journal:	Human Mutation
Manuscript ID	humu-2018-0045
Wiley - Manuscript type:	Research Article
Date Submitted by the Author:	29-Jan-2018
Complete List of Authors:	geoffroy, veronique; Laboratoire de Génétique Médicale, Institut de Génétique Médicale d'Alsace, INSERM U1112, Fédération de Médecine Translationnelle de Strasbourg (FMTS), Université de Strasbourg Stoetzel, Corinne; UMR_S INSERM U1112, IGMA, Laboratoire de Génétique médicale, Scheidecker, Sophie; UMR_S INSERM U1112, IGMA, Laboratoire de Génétique médicale, SCHAEFER, Elise; Hôpital de Hautepierre, GENETICS PERRAULT, Isabelle; INSERM U781, GENETICS Bär, Séverine; Department of Molecular and Cellular Genetics, UMR7156, Centre National de Recherche Scientifique (CNRS), Université de Strasbourg, Strasbourg, France Kröll, Ariane; UMR_S INSERM U1112, IGMA, Laboratoire de Génétique médicale, Delbarre, Marion; Hôpitaux universitaire de Strasbourg, Laboratoires de diagnostic génétique Antin, Manuela; Nouvel Hopital Civi, Laboratoire de Diagnostic Génétique Jaeger, Anne-Sophie; Hôpitaux universitaire de Strasbourg, Laboratoires de diagnostic génétique Henry, Charline; Inserm U1163-institut Imagine Blanché, Hélène; Fondation Jean-Dausset-Centre d'Etude du Polymorphisme Humain (CEPH), Institut de Génétique Moléculaire, Decker, Eva; Bioscentia, Kloth, Katja; Institut für Humangenetik, Universitätsklinikum Hamburg- Eppendorf, Hamburg, Germany Klaus, Günter; University Marburg, KfH-Nierenzentrum für Kinder und Jugendliche, Marburg, Germany Mache, Christopher; Graz, Martin-Coignard, dominique; CH le Mans, Service de Génétique Médicale McGinn, Steven; CNRGH, Institut de Biologie François Jacob, DRF, CEA, Evry, France Boland, Anne; CNG Deleuze, Jean-François; CNG Friant, Sylvie; Department of Molecular and Cellular Genetics, UMR7156, Centre National de Recherche Scientifique (CNRS), Université de Strasbourg, Strasbourg, France SAUNIER, Sophie; Université René Descartes, ; INSERM U-574, Hopital Necker, Enfants Malades

1		
2 3		
4		ROZET, Jean-Michel; INSERM U781, GENETICS Bergmann, Carsten: Aachen University, Department of Human Genetics
5		Dollfus, Helene; UMR_S INSERM U1112, IGMA, Laboratoire de Génétique
6 7		médicale, Mullar, Japan Hânitaux universitaire de Strasbeurg, Laborateires de
7 8		diagnostic génétique: Laboratoire de Génétique Médicale, Institut de
9		Génétique Médicale d'Alsace, INSERM U1112, Fédération de Médecine
10		Translationnelle de Strasbourg (FMTS), Université de Strasbourg,
11		Mainzer-Saldino syndrome, IFT140, Structural variation, tandem
12	Key Words:	duplication, whole-genome sequencing, Alu-mediated recombination, copy
13		
15		
16	1	
17		
18 19		Manuscripts
20		
21		
22		
23		
24		
26		
27		
28		
30		
31		
32		
33		
35		
36		
37		
38 30		
40		
41		
42		
43 44		
45		
46		
47		
48		
49 50		
51		
52		
53		
54 55		
56		
57		
58		
59 60		John Wiley & Sons, Inc.
		-

Whole Genome Sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in *IFT140*

Véronique Geoffroy^{1,*}, Corinne Stoetzel^{1,*}, Sophie Scheidecker^{1,2}, Elise Schaefer^{1,3}, Isabelle Perrault⁴, Séverine Bär⁵, Ariane Kröll¹, Marion Delbarre², Manuela Antin², Anne-Sophie Leuvrey², Charline Henry⁶, Hélène Blanché⁷, Eva Decker⁸, Katja Kloth⁹, Günter Klaus¹⁰, Christoph Mache¹¹, Dominique Martin-Coignard¹², Steven McGinn¹³, Anne Boland¹³, Jean-François Deleuze^{7,13}, Sylvie Friant⁵, Sophie Saunier⁶, Jean-Michel Rozet⁴, Carsten Bergmann^{8,14}, Hélène Dollfus^{1,15}, Jean Muller^{1,2}

¹Laboratoire de Génétique médicale, UMR_S INSERM U1112, IGMA, Faculté de Médecine FMTS, Université de Strasbourg, Strasbourg, France

²Laboratoires de Diagnostic Génétique, Hôpitaux Universitaires de Strasbourg, Strasbourg Cedex, France

³Service de Génétique Médicale, Hôpitaux Universitaires de Strasbourg, Strasbourg, France.

⁴Laboratory of Genetics in Ophthalmology (LGO), INSERM UMR1163, Institute of Genetic Diseases, Imagine, Paris Descartes University, 75015 Paris, France.

⁵Department of Molecular and Cellular Genetics, UMR7156, Centre National de Recherche

Scientifique (CNRS), Université de Strasbourg, Strasbourg, France.

⁶INSERM, U983, Paris Descartes University, Paris, France

⁷Centre d'études du polymorphisme humain-Fondation Jean Dausset, Paris, France

⁸Center for Human Genetics, Bioscientia, Ingelheim, Germany

⁹Institut für Humangenetik, Universitätsklinikum Hamburg-Eppendorf, Hamburg, Germany

¹⁰University Marburg, KfH-Nierenzentrum für Kinder und Jugendliche, Marburg, Germany

¹¹Department of Pediatrics, Medical University of Graz, Graz, Austria.

Human Mutation

¹²Service de Génétique, Centre Hospitalier, CCLAD, Le Mans, France
¹³CNRGH, Institut de Biologie François Jacob, DRF, CEA, Evry, France
¹⁴Department of Medicine, University Hospital Freiburg, Freiburg, Germany
¹⁵Centre de Référence pour les affections rares en génétique ophtalmologique, CARGO, Filière SENSGENE, Hôpitaux Universitaires de Strasbourg, 67091 Strasbourg, France.

*equal contributor

Corresponding Author: Jean Muller

Email address: jeanmuller@unistra.fr

Abstract

Ciliopathies represent a wide spectrum of rare diseases with overlapping phenotypes and a high genetic heterogeneity. Among those, *IFT140* is implicated in a variety of phenotypes ranging from isolated retinis pigmentosa to more syndromic cases such as the Bardet-Biedl syndrome. Using whole genome sequencing in patients with uncharacterized ciliopathies, we identified a novel recurrent tandem duplication of exon 27 to 30 (6.7 kb) in *IFT140*, c.3454-488_4182+2588dup p.(Tyr1152_Thr1394dup), missed by whole exome sequencing. Pathogenicity of the mutation was assessed on the patients' skin fibroblasts. Several hundreds of patients with a ciliopathy phenotype were screened and biallelic mutations were identified in 11 families representing 12 pathogenic variants of which 7 are novel. Among those unrelated families especially with a Mainzer-Saldino syndrome, 8 carried the same tandem duplication (2 at the homozygous state and 6 at the heterozygous state).

In conclusion, we demonstrated the implication of structural variations in *IFT140* related diseases expanding its mutation spectrum. We also provide evidences for a unique genomic event mediated by an *Alu-Alu* recombination occurring on a shared haplotype. We confirm that whole genome sequencing can be instrumental in the ability to detect structural variants for genomic disorders.

Keywords: *IFT140*, Mainzer-Saldino syndrome, structural variation, copy number variation, tandem duplication, *Alu*-mediated recombination, whole-genome sequencing.

Background

Mainzer-Saldino syndrome (MSS, MIM 266920) is a rare (< 1/1,000,000) autosomal recessive ciliopathy characterized by severe early-onset retinal dystrophy, phalangeal cone-shaped epiphyses, chronic renal failure, and mild radiographic abnormality of the proximal femur (Perrault, et al., 2012), known to be caused by *IFT140* (Khan, et al., 2014; Perrault, et al., 2012; Schmidts, et al., 2013) and *IFT172* mutations (Halbritter, et al., 2013). Both genes have been implicated in several other ciliopathies ranging from isolated Retinis Pigmentosa (RP) to more syndromic cases such as Jeune or Bardet-Biedl syndromes (Bifari, et al., 2016; Bujakowska, et al., 2015; Schaefer, et al., 2016).

Intraflagellar transport (*IFT*) genes are involved in a bidirectional (anterograde and retrograde) transport process essential for the assembly and the maintenance of the cilia through the redistribution of ciliary proteins between the cell body and the cilium. In particular, IFT140 a component of the IFT complex A (IFT-A) responsible of the retrograde IFT, is vital for both the development and the maintenance of outer segments of photoreceptors and has a specific role in opsin transport across the connecting cilium (Blacque, et al., 2006; Crouse, et al., 2014) of the photoreceptors. Previously, 46 different mutations in 56 families have been reported in *IFT140* (Additional File 1: Table S1), encompassing missenses, essential splice sites, stop and frameshifts mutations but no structural variations (SV) such as copy number variant (CNV). Interestingly, 10 mono allelic variants cases have also been reported with no second pathogenic allele detected to date (Additional File 1: Table S1).

In this study, we report for the first time a structural variation (tandem duplication) in *IFT140* identified by Whole Genome Sequencing (WGS) and missed by Whole Exome Sequencing (WES). The impact of this mutation was further assessed in patients' skin fibroblasts. Given

the difficulty to identify this mutation and the genomic context surrounding the breakpoints, we speculated that it might have been missed by other genetic screenings and that several other families might carry this mutation. We finally report 8 unrelated families carrying the mutation either at the homozygous or the heterozygous state, out of 11 families identified with *IFT140* biallelic mutations in total. Moreover, the characterization of the breakpoints allowed to delineate a potential molecular mechanism and to design a specific duplex PCR that will help screening further patients (including our cohorts).

Materials and Methods

Subjects

Study protocols used in each cohort have been approved by the corresponding Institutional Review Board or equivalent committees (as an example in Strasbourg, "Comité Protection des Personnes" EST IV, N°DC-20142222), and written informed consent was given from each participant or parents. Our research complies with the Declaration of Helsinki. Written informed consent for open-access publication was provided by the participants or their parents. DNA of additional affected and unaffected family members was requested whenever it was considered informative. Skin fibroblasts were obtained for family A. Clinical data for all 11 families are presented in Additional File 1: Table S2.

Whole Genome Sequencing

WGS was performed for the two affected siblings from family A (A-II.1 and A-II.2) and their parents (A-I.1 and A-I.2) by the Centre National de Génotypage (Institut de Génomique, CEA). Genomic DNA was used to prepare a library for whole genome sequencing, using the Illumina TruSeq DNA PCR-Free Library Preparation Kit, according to the manufacturer's

Human Mutation

instructions. After normalization and quality control, qualified libraries have been sequenced on a HiSeq2000 platform from Illumina (Illumina Inc., CA, USA), as paired-end 100 bp reads. At least 3 lanes of HiSeq2000 flow cell have been produced for each sample, in order to reach an average sequencing depth of 30x for each sample. Sequence quality parameters have been assessed throughout the sequencing run and standard bioinformatics analysis of sequencing data was based on the Illumina pipeline to generate FASTQ file for each sample. The sequence reads were aligned to the reference sequence of the human genome (GRCh37) using the Burrows-Wheeler Aligner (BWA V7.12) (Li and Durbin, 2010). The UnifiedGenotyper and HaplotypeCaller modules of the Genome Analysis ToolKit (GATK) (DePristo, et al., 2011), Platypus (http://www.well.ox.ac.uk/platypus) and Samtools (Li, et al., 2009) were used for calling both single nucleotide variations (SNV) and small insertion/deletion (indel).

Bioinformatics analysis

Annotation and ranking of SNV and indel were performed by VaRank (Geoffroy, et al., 2015) in combination with the Alamut Batch software (Interactive Biosoftware, Rouen, France). Very stringent filtering criteria were used for excluding non-pathogenic variants, in particular: (1) variants represented with an allele frequency of more than 1% in public variation databases either dbSNP 138 (Sherry, et al., 2001), the Exome Variant Server (NHLBI GO Exome Sequencing Project, http://evs.gs.washington.edu/EVS/), the 1000Genomes (Genomes Project, et al., 2015), the ExAC browser database(Lek, et al., 2016) or our internal exome database, (2) variants in 5' UTR, 3' UTR, downstream, upstream or intronic locations without pathogenic prediction of local splice effect, and (3) synonymous variants without prediction of local splice effect on the nearest splice site was predicted using MaxEntScan (Yeo and Burge, 2004), NNSplice (Reese, et al., 1997) and Splice Site Finder (Shapiro and

Senapathy, 1987). Our analysis was focused on compound heterozygous and homozygous variants (SNV/indel/SV) consistent with a recessive mode of transmission. Structural variants were predicted using by default the CANOES program (Backenroth, et al., 2014) and annotated thanks to our in house script AnnotSV (manuscript in preparation, http://www.lbgi.fr/AnnotSV/) based on the classical annotations such as the Database of Genomic Variants (DGV) (MacDonald, et al., 2014). The *IFT140* nomenclature is based on the accession number NM_014714.3 from the RefSeq database (O'Leary, et al., 2016). Genomic coordinates are defined according to GRCh37/hg19 assembly downloaded from the University of California Santa Cruz (UCSC) genome browser (Tyner, et al., 2017).

Sanger validation and segregation

Sanger sequencing was performed by PCR amplification with 50 ng of genomic DNA template. The primers were designed with Primer 3 (<u>http://frodo.wi.mit.edu/primer3</u>) and are detailed in Additional File 1: Table S3. Bidirectional sequencing of the purified PCR products was performed by GATC Sequencing Facilities (Konstanz, Germany).

qPCR quantification

Absolute quantification was performed using the SyberGreen Mastermix Quantitect (Qiagen) measured on the LightCycler 480 (Roche). Amplicons were designed in exon 30 of *IFT140* and compared to two reference genes (*HBB* and *HMBS*). Each samples have been done in duplicate and standard deviation was <0.2. Standard curve have been established using 5 different DNA quantity (50ng, 25ng, 10ng, 5ng and 1ng) and PCR efficiency was assessed for each amplicon (1.9<PCR efficiency<2.0). Ratio between the mean concentrations from the amplicon of interest compared to reference gene was collected. The mean values for both

Human Mutation

control genes was calculated for each patient and plotted. The primers were designed with Primer 3 and are detailed in Additional File 1: Table S3

RNA analysis

RNA was extracted from skin fibroblasts of individual II.1 and a healthy unrelated control using Rneasy RNA kit (Qiagen) then we performed reverse transcription using the iScriptTM cDNA Synthesis Kit (BioRad, Hercules, CA).

Cell culture

Fibroblasts of patients and control individuals were obtained by skin biopsy as previously described (Scheidecker, et al., 2014). To induce primary cilium formation, cells were deprived of serum by growth for 24 hrs in DMEM with 1% PSG but only 0.1% FCS (conditions -FCS) as previously described (Stoetzel, et al., 2016).

Immunofluorescence

Primary fibroblasts from patients and control individuals were grown in Nunc Lab-Tek chamber slides (Thermo Scientific) and ciliogenesis was done as described above. Primary cilia were labelled with an antibody directed against acetylated a-tubulin highlighting the axoneme41. Pictures were taken either on a fluorescence microscope (Figure 2A) or a confocal microscope (Figure 2C). Primary and secondary antibodies used in this study as well as their dilution are indicated in Additional File 1: Table S4.

Cohort screening

A duplex PCR was designed to specifically detect the tandem duplication. Primers and example conditions are provided in Additional File 1: Table S3 and Figure S1. Additional

targeted exome sequencing datasets have been reanalyzed (see Additional File 1: supplementary Methods).

Alu pair analysis

We obtained the annotations and sequences for the two *Alu* repeat in the duplication junction region from the RepeatMasker track of the UCSC genome browser (Tyner, et al., 2017). We subsequently aligned the *Alu* sequences and computed the identity using the online tool EMBOSS Matcher (Rice, et al., 2000).

Results

Identification of biallelic mutations in IFT140

Two affected individuals born from healthy non consanguineous parents (family A, Figure 1A), named II.1 and II.2, were referred to our lab with retinitis pigmentosa, short stature (152cm and 158cm respectively at 16 and 15yo), brachydactyly, moderate renal failure and overweight for II.1 (Additional File 1: Table S2). This Bardet-Biedl like phenotype prompted us to perform an extensive first genetic analysis including Sanger sequencing of recurrent BBS mutations (Muller, et al., 2010), targeted exome sequencing (Redin, et al., 2012) and WES without success. Indeed no biallelic mutations including copy number variant could be identified (see Additional File 1: Table S5). Hence, considering that affected individuals from family A may harbor mutations in a region not (well) covered by the WES (i.e. deep intronic, promotor...), we applied WGS to the 2 affected individuals and their parents. A combined analysis including both SNV/indel and structural variant revealed biallelic mutations in *IFT140*, a gene known to be responsible of several ciliopathies (ranging from isolated RP to more syndromic cases such as Jeune or Mainzer-Saldino syndromes) compatible with the phenotype of the affected patients.

First, the two affected individuals were found heterozygous for a possible splice mutation in intron 20, c.2577+25G>A, predicted to have a local splice effect as a new strong donor site. This variant was reported previously and is not present in any variation database (e.g. ExAC or 1000G). Co-segregation analysis revealed the paternal inheritance (Figure 1A, primers used are detailed in Additional File 1: Table S3). Patient's RNA analysis confirmed that a novel donor splice site within the intron 20 resulted in the incorporation of 21 additional bp (r.2577_2578insGTGAGGGGCGCCCGCCATGGG) that are predicted to add 7 new amino acids to the protein sequence (p.Leu859 Glu860insValArgGlyAlaArgHisGly) (Figure 1B).

Second, in *trans* to this mutation, structural variant analysis revealed a maternally inherited (Figure 1A) duplication of exons 27 to 30 (of sizes 6.7 kb). Analysis of the mapped reads revealed that the duplication is occurring in tandem in direct orientation within the gene (Additional File 1: Figure S2) with breakpoints located in intron 26 and intron 30. This tandem duplication has not been reported previously in patients and is not present in DGV. The results were confirmed by qPCR (Figure 1C) delineating the expected maternal inheritance. The duplication is predicted to be in frame and to add 243 amino acids within the tetratricopeptide repeat (TPR) domain of the IFT140 protein (c.3454-488_4182+2588dup, p.Tyr1152_Thr1394dup). RNA analysis revealed that the duplicated exons are transcribed (Figure 1D). Western blot analysis revealed only a single band of the approximate size of the wild type protein suggesting that the allele carrying the duplication is not detected as a protein (Figure 1E).

Immunofluorescence analysis in patients fibroblasts revealed that IFT140 is mislocalized compared to sex/age matched control cells. Indeed, when grown in ciliary conditions, patient skin fibroblasts developed primary cilia less often (Figure 2A). Thus while 75% of control cells developed a primary cilium, only about 50-55% of patient cells did so (Figure 2B). It is to notice that primary cilia did not seem to be altered since when cells were deprived of

serum, fixed for immunofluorescence and labeled with an anti-acetylated tubulin antibody, their cilium as observed by fluorescence microscope and measured with image J (Schneider, et al., 2012) was about 2-2,5 μ m both in control and patient cells (Figure 2**B**). Nevertheless, a difference in organisation could be noticed when co-labelling was performed with an anti-IFT140 antibodies (Figure 2**C**). Then, localisation of IFT140 at the base of the cilium could be observed using confocal microscopy in 82% of ciliated control cells but only in about 14% of ciliated patient fibroblasts (Figure 2**D**). Thus, the mutations found in the patients lead to a significant decrease of cilium formation and a loss of IFT140 localisation at the base of the cilium.

Mutation screening in a large cohort

To explore additional cases of this duplication possibly missed by prior analysis, we retrospectively screened our cohorts and reanalyzed available high throughput sequencing panels including the *IFT140* gene. As mutations in *IFT140* are known to cause isolated to syndromic retinal degeneration (Bifari, et al., 2016), this included 126 patients using the Leber panel of the Imagine institute, 117 patients using the RP panel of Strasbourg Hospital , and 104 patients using the Ciliome panel of the Imagine institute (see Methods and Additional File 1: methods online). We also setup a duplex PCR around the breakpoints (Additional File 1: Figure S1) and screened all our negative cases, including 207 from our Bardet-Biedl cohort, 150 isolated RP and 40 *IFT140* heterozygous patients. All together we identified 11 additional families positive for biallelic mutations in *IFT140* (Figure 3 and Additional File 1: Table S1) among which 10 families have a phenotype compatible with a Mainzer Saldino syndrome and one patient presents with isolated retinitis pigmentosa. Among the MSS patients, three patients presented a phenotype overlapping with another ciliopathy: two with Jeune syndrome and one with Sensenbrenner syndrome. Eight of the 11 families carried the exact same tandem duplication either at the heterozygous state (3 copies) or at the

John Wiley & Sons, Inc.

Human Mutation

homozygous state (4 copies) (Figure 3A, Additional File 1: Figure S3). Among the novel mutations identified, we observed 2 missenses at the heterozygous state c.1319T>C (p.Leu440Pro), c.2177C>T (p.Pro726Leu) that are both predicted deleterious (Additional File 1: supplementary Methods). Among the novel mutations we also identified another CNV, a large deletion encompassing exon 27 to 29 (c.3454-1005_4040+737delinsCCC, Additional File 1: Figure S4) that does not share the same breakpoints compared to the tandem duplication. Thus to date, 120 pathogenic alleles have been reported for *IFT140* of which the tandem duplication represents 10 alleles. Interestingly this tandem duplication is the second most frequent mutation for *IFT140*-affected patients.

Founder effect or recurrent mechanism?

Given the relatively high frequency (8/65 families) of this duplication in the patients described with *IFT140* mutations, we performed 3 additional explorations to assess whether the duplication was an independent recurrent mechanism or the same mutational event derived from a common ancestry suggesting a founder effect. First, we screened a French cohort of 354 WGS of healthy individuals (FREX dataset), for which CNV analysis (CANOES) as well as visual inspections of the barn files (IGV batch scripting (Thorvaldsdottir, et al., 2013)) did not reveal any corresponding allele, as expected due to the *IFT140* diseases prevalence (e.g. MSS: 1/1,000,000). Second, we compared the haplotype at the *IFT140* locus from the different patients for which NGS data was available. In total 15 haplotypes, including 6 carrying the duplication, from 3 different families, were analyzed (see Additional File 1: Table S6). We observed a shared haplotype between the duplication carriers at the *IFT140* locus, suggesting a founder effect. Third, we characterized the genomic localization around the breakpoint junctions. Sanger sequencing confirmed the breakpoint's coordinates (Figure 1A and Additional File 1: Figure S2) and demonstrated that the exact same breakpoint is shared among all patients tested (Family C was not investigated). The

breakpoints overlapped two distinct repeated elements from the *Alu* family, *AluJb* and *AluJr* (Figure 4A). Pairwise alignment of the two *Alu* sequences (Additional File 1: Figure S5) revealed a conservation of 68% of identity that reached 81% in the junction region (Figure 4B). The tandem duplication is resulting in an *Alu* hybrid (Figure 4C) at both sides. *Alu-Alu* recombination is a prominent mechanism underlying the formation of pathogenic SV associated with distinct diseases (Boone, et al., 2014; Stankiewicz, et al., 2009). Altogether, this led us to speculate that this tandem duplication is a rare event, which arises from a common ancestor due to an *Alu-Alu* mediated genomic recombination.

Discussion

The advent of next generation sequencing, especially Whole Exome Sequencing (WES), in the past 10 years was a tremendous step that led to the identification of many mutations accelerating the discovery of novel genes involved in human genetic diseases (Chong, et al., 2015). Nevertheless, the diagnostic yield of WES is plateauing between 25% to 50% depending on the disease (Chong, et al., 2015; Taylor, et al., 2015) leaving many patients still with no molecular diagnosis. The use of RNA sequencing has recently extended the percentage by 10% to 66% (depending on the context: with or without a strong candidate by prior DNA sequencing) by helping identifying unseen defects or interpreting variations found in the WES (Cummings, et al., 2017; Kremer, et al., 2017). Lastly, Whole Genome Sequencing (WGS) of human patients has proven to be an instrumental tool for identifying the whole spectrum of genetic anomalies (Gilissen, et al., 2014) and will replace in a close future other genetic screening.

In line with this, we applied a range of genetic screening including targeted sequencing, WES and then finally WGS to unsolved patients affected with a ciliopathy. The combination of

Human Mutation

WGS and CNV analysis was able to uncover a tandem duplication unseen by the WES and to reconsider a distant splice site mutation.

Interestingly, the tandem duplication could be identified in family A only by using the WGS and not the WES data. Indeed, CANOES failed to identify the event in one of the individuals (Additional File 1: Table S7), which prevented us from considering this as a mutation of interest but made us rather discard it as a false positive of the method. Thereupon, it is well known that CNV detection from next generation sequencing (NGS) datasets is feasible and many publication reported positive results either on gene panels or WES (de Ligt, et al., 2013; Redin, et al., 2012). However, this remains challenging (Tan, et al., 2014). WGS provides a much more uniform distribution of sequencing-quality parameters and by nature does not have the restriction of noncontiguous regions of interests (captured exons). Therefore, it is more suited for SV identification (Belkadi, et al., 2015). Having that in mind, we further tested the CNV detection using 2 other popular programs (see Additional File 1: Table S7) which failed to detect the tandem duplication in family A patients (Additional File 1: Table S7). These results confirmed many efforts are still required to ensure a high quality CNV detection from NGS data, even with the large number of programs and methods available. Nevertheless, thanks to the split reads from WGS data, once a CNV is detected one can easily define the CNV breakpoints and further characterize the mechanism, as done in this study (Additional File 1: Figure S2).

Analysis of the breakpoint positions and in particular the split reads allowed us to identify overlapping *Alu* elements at the breakpoint junction. Alignment of the *Alu* regions surrounding the junction revealed that both elements share 81% identity and exhibit a microhomology region of 6 nucleotides. Because the recombination occurred between homologous sequences (e.g. imperfect match substrates), it is most likely mediated by mechanisms other than nonallelic homologous recombination, which are mostly recurrent

events (NAHR, for review see (Carvalho and Lupski, 2016)). Among the possible molecular mechanisms, that include non-homologous end joining (NHEJ) and replication-based mechanisms such as break-induced replication (BIR), microhomology-mediated BIR (MMBIR), serial replication slippage (SRS) and fork stalling and template switching (FoSTeS), we believe that the presence of a microhomology region points most likely to a MMBIR driven recombination. Given the nonrecurrent nature of those mechanisms, the common haplotype and the exact same breakpoint junction observed in most of our patients, we hypothesized that the genomic event that gave rise to this tandem duplication occurred once on a common *IFT140* haplotype.

IFT140 is a 1462-amino-acid protein encoded by 31 exons and composed of 5 WD repeats and 9 tetratricopeptide repeats (TPR) both known to act as protein-protein interaction domains that might be involved in IFT140 interactions within the IFT-A complex (Zhu, et al., 2017). To date, 45 different pathogenic alleles have already been described throughout the *IFT140* gene with no clustering or domain preference (Figure 4A). Most of them are private mutations but several are found multiple times like the c.634G>A (p.Gly212Arg) (7 alleles) which has been proven to affect the splicing of exon 6 (Helm, et al., 2017) or the c.2399+1G>T (5 alleles) (Additional File 1: Table S1). The most frequent mutation (31 alleles) is a missense (c.1990G>A, p.Glu664Lys) that has been observed in multiple studies and especially in 11 consanguineous families from the Arabian Peninsula sharing a common ancestor (Bifari, et al., 2016) which might bias the allele count. Nevertheless, the tandem duplication described in our study is the second most frequent cause of mutation in *IFT140* representing 10 alleles. In family A, the two mutations identified required extensive and careful analysis to be considered. One mutation was predicted to affect the splicing of exon 20 (c.2577+25G>A)

and suggested to add 7 amino acid to the protein sequence

which was confirmed at the RNA level (r.2577 2578insGTGAGGGGGGCGCCCGCCATGGG),

(p.Leu859_Glu860insValArgGlyAlaArgHisGly). The tandem duplication of exons 27 to 30 (c.3454-488_4182+2588dup, p.Tyr1152_Thr1394dup) is predicted to add 243 amino acids within the TPR repeat domain. Skin fibroblast analysis revealed RNA synthesis in the patient cells but no protein could be identified by Western Blot at the predicted size. We hypothesize that the protein was not stable enough to maintain its 3D structure. Functional analysis on the patients' cells further revealed a reduced level of ciliated cells and mislocalization of the IFT140 mutant away from the cilia base.

The Mainzer Saldino syndrome is a syndrome characterized by skeletal phenotype with phalangeal cone-shaped epiphyses, chronic renal disease, and retinal dystrophy. Overlapping phenotypes have been described with other skeletal ciliopathies such as the Jeune or Sensenbrenner syndrome. In our cohort the diagnosis of MSS was not made in all patients before the molecular analysis. However, all of them except one with an isolated retinitis pigmentosa, fulfilled the criteria for this diagnosis after a new careful clinical and radiological examination. Indeed, 11 patients presented digits anomalies, including 8 with phalangeal cone-shaped epiphyses. Nine patients developed renal failure among which 7 have a severe and early onset renal disease (1.5 years old to 23 years old) as previously described (Schmidts, et al., 2013). The retinal dystrophy is constantly reported with a highly variable age of onset. Among our cohort, a single patient presented an isolated retinitis pigmentosa (as previously reported by others (Hull, et al., 2016)). Two patients have thoracic dystrophy which is a clinical criteria of Jeune syndrome and one patient has a craniosynostosis compatible with a Sensenbrenner syndrome. These descriptions illustrate well the overlap existing between the different ciliopathies associated with mutation in IFT140. On a molecular level, no genotype-phenotype correlation could be established in our cohort, especially regarding the patients carrying the duplication at the homozygous or the heterozygous state.

Conclusions

In summary, we report here 11 novel unrelated families with mutations in *IFT140*. Among them, 8 families carry a recurrent tandem duplication of 4 exons either at the heterozygous state or the homozygous state, for which we have assessed the pathogenicity in the pateints cells. This is the first time that a structural variation is reported in *IFT140* expanding the mutation spectrum for this gene. Notably, this large duplication was missed by the WES analysis but uncoverd thanks to the whole genome, pointing out the power of such analysis.

Availability of data and material

Data generated or analyzed during this study are included in the published article and the corresponding supplementary data. The raw sequencing data generated in the course of this study are not publicly available due to the protocol and the corresponding consents used that did not include such information. All variants have been submitted to ClinVar (https://www.ncbi.nlm.nih.gov/clinvar/). Anonymised NGS data and genomic variant data files will be made available upon request from qualified investigators studying the molecular basis of genomic disorders. Datasets can be obtained via the corresponding author on reasonable request.

Acknowledgements

We would like to thank the patients and their family for their participation. We thank Daniel Backenroth and Olivier Quenez for their help in the bioinformatics setup of CANOES, Arnaud Kress and Antony Le Béchec for informatic support, Amélie Piton, Bénédicte Gérard and Ilia Humbert for scientific discussion. We also thank Emmanuelle Génin, Pierre Lindenbaum and Richard Redon for giving us access to the genome dataset from the FREX

Human Mutation

project and the Genomic and Bioinformatic Platform of Institut Imagine (Patrick Nitschké, and Cécile Masson).

Disclosure Statment

Eva Decker and Carsten Bergmann are employees of Bioscientia/Sonic Healthcare. Günter Klaus benefited from a travel grants from The Apheresis Research Institute Cologne, the DGFF (Die Deutsche Gesellschaft zur Bekämpfung von Fettstoffwechselstörungen und ihren Folgeerkrankungen) and is consultant to Vifor pharma, Switzerland concerning a pediatric phosphat binder trial.

Funding Information

The WGS research was supported by the Laboratory of Excellence GENMED (Medical Genomics) grant no. ANR-10-LABX-0013 managed by the National Research Agency (ANR) part of the Investment for the Future program. Whole exome sequencing was performed by the IGBMC Microarray and Sequencing platform, a member of the 'France Génomique' consortium (ANR-10-INBS-0009) and funded by "La Fondation Maladie Rare". The RP panel is supported by the French program PHRC I 2013 HUS N° 5724 and by grants from the Retina France to IP, UNADEV- AVIESAN ITMO MNP to JMR. In addition, CB holds a part-time faculty appointment at the University of Freiburg. His research lab receives support from the Deutsche Forschungsgemeinschaft (DFG) Collaborative Research Centre (SFB) KIDGEM 1140 and the Federal Ministry of Education and Research (BMBF, 01GM1515C).

References

 Backenroth D, Homsy J, Murillo LR, Glessner J, Lin E, Brueckner M, Lifton R, Goldmuntz E, Chung WK, Shen Y. 2014. CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Res 42(12):e97.

Belkadi A, Bolze A, Itan Y, Cobat A, Vincent QB, Antipenko A, Shang L, Boisson B, Casanova JL, Abel L. 2015. Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. Proc Natl Acad Sci U S A 112(17):5473-8.

Bifari IN, Elkhamary SM, Bolz HJ, Khan AO. 2016. The ophthalmic phenotype of IFT140related ciliopathy ranges from isolated to syndromic congenital retinal dystrophy. Br J Ophthalmol 100(6):829-33.

Blacque OE, Li C, Inglis PN, Esmail MA, Ou G, Mah AK, Baillie DL, Scholey JM, Leroux MR. 2006. The WD repeat-containing protein IFTA-1 is required for retrograde intraflagellar transport. Mol Biol Cell 17(12):5053-62.

Boone PM, Yuan B, Campbell IM, Scull JC, Withers MA, Baggett BC, Beck CR, Shaw CJ, Stankiewicz P, Moretti P and others. 2014. The Alu-rich genomic architecture of SPAST predisposes to diverse and functionally distinct disease-associated CNV alleles. Am J Hum Genet 95(2):143-61.

Bujakowska KM, Zhang Q, Siemiatkowska AM, Liu Q, Place E, Falk MJ, Consugar M, Lancelot ME, Antonio A, Lonjou C and others. 2015. Mutations in IFT172 cause isolated retinal degeneration and Bardet-Biedl syndrome. Hum Mol Genet 24(1):230-42.

Carvalho CM, Lupski JR. 2016. Mechanisms underlying structural variant formation in genomic disorders. Nat Rev Genet 17(4):224-38.

Chong JX, Buckingham KJ, Jhangiani SN, Boehm C, Sobreira N, Smith JD, Harrell TM, McMillin MJ, Wiszniewski W, Gambin T and others. 2015. The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. Am J Hum Genet 97(2):199-215.

Crouse JA, Lopes VS, Sanagustin JT, Keady BT, Williams DS, Pazour GJ. 2014. Distinct functions for IFT140 and IFT20 in opsin transport. Cytoskeleton (Hoboken) 71(5):302-10.

Cummings BB, Marshall JL, Tukiainen T, Lek M, Donkervoort S, Foley AR, Bolduc V, Waddell LB, Sandaradura SA, O'Grady GL and others. 2017. Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Sci Transl Med 9(386).

de Ligt J, Boone PM, Pfundt R, Vissers LE, Richmond T, Geoghegan J, O'Moore K, de Leeuw N, Shaw C, Brunner HG and others. 2013. Detection of clinically relevant copy number variants with whole-exome sequencing. Hum Mutat 34(10):1439-48.

DePristo MA, Banks E, Poplin R, Garimella KV, Maguire JR, Hartl C, Philippakis AA, del Angel G, Rivas MA, Hanna M and others. 2011. A framework for variation discovery and genotyping using next-generation DNA sequencing data. Nat Genet 43(5):491-8.

Genomes Project C, Auton A, Brooks LD, Durbin RM, Garrison EP, Kang HM, Korbel JO, Marchini JL, McCarthy S, McVean GA and others. 2015. A global reference for human genetic variation. Nature 526(7571):68-74.

Geoffroy V, Pizot C, Redin C, Piton A, Vasli N, Stoetzel C, Blavier A, Laporte J, Muller J. 2015. VaRank: a simple and powerful tool for ranking genetic variants. PeerJ 3:e796.

Gilissen C, Hehir-Kwa JY, Thung DT, van de Vorst M, van Bon BW, Willemsen MH, Kwint M, Janssen IM, Hoischen A, Schenck A and others. 2014. Genome sequencing identifies major causes of severe intellectual disability. Nature 511(7509):344-7.

Halbritter J, Bizet AA, Schmidts M, Porath JD, Braun DA, Gee HY, McInerney-Leo AM, Krug P, Filhol E, Davis EE and others. 2013. Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. Am J Hum Genet 93(5):915-25.

1	
2	
3	Helm BM, Willer JR, Sadeghpour A, Golzio C, Crouch E, Vergano SS, Katsanis N, Davis
4	EE. 2017. Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic
5	mutation in IFT140 that causes Mainzer-Saldino syndrome. Hum Genomics 11(1):16.
6	Hull S, Owen N, Islam F, Tracey-White D, Plagnol V, Holder GE, Michaelides M, Carss K,
7	Raymond FL, Rozet JM and others. 2016. Nonsyndromic Retinal Dystrophy due to Bi-Allelic
8	Mutations in the Ciliary Transport Gene IFT140. Invest Ophthalmol Vis Sci 57(3):1053-62.
9	Khan AO, Bolz HJ, Bergmann C. 2014. Early-onset severe retinal dystrophy as the initial
10	presentation of IFT140-related skeletal ciliopathy. J AAPOS 18(2):203-5.
11	Kremer LS, Bader DM, Mertes C, Kopajtich R, Pichler G, Juso A, Haack TB, Graf E,
12	Schwarzmayr T. Terrile C and others. 2017. Genetic diagnosis of Mendelian disorders via
13	RNA sequencing. Nat Commun 8:15824.
14	Lek M Karczewski KI Minikel EV Samocha KE Banks E Fennell T O'Donnell-Luria AH
15	Ware IS Hill AI Cummings BB and others 2016 Analysis of protein-coding genetic
16	variation in 60 706 humans. Nature 536(7616):285 01
/ 10	Li H Durbin P 2010 Fast and accurate long read alignment with Burrows Wheeler
10	transform Diginformation 26(5):580.05
19	Li II Handaalaan D. Waaalaan A. Fannall T. Duan I. Haman N. Marth C. Ahaaasia C. Durhin
20	Li n, nanusakei D, wysokei A, rennen I, Kuan J, nomer N, Marin G, Abecasis G, Durbin D, Canoma Draigat Data Dragoning S, 2000. The Construct Alignment/Mark C, and the Second S, 2000.
21	R, Genome Project Data Processing S. 2009. The Sequence Alignment/Map format and
22	SAMtools. Bioinformatics 25(16):20/8-9.
23	MacDonald JR, Ziman R, Yuen RK, Feuk L, Scherer SW. 2014. The Database of Genomic
25	Variants: a curated collection of structural variation in the human genome. Nucleic Acids Res
26	42(Database issue):D986-92.
27	Muller J, Stoetzel C, Vincent MC, Leitch CC, Laurier V, Danse JM, Helle S, Marion V,
28	Bennouna-Greene V, Vicaire S and others. 2010. Identification of 28 novel mutations in the
29	Bardet-Biedl syndrome genes: the burden of private mutations in an extensively
30	heterogeneous disease. Hum Genet 127(5):583-93.
31	O'Leary NA, Wright MW, Brister JR, Ciufo S, Haddad D, McVeigh R, Rajput B, Robbertse
32	B, Smith-White B, Ako-Adjei D and others. 2016. Reference sequence (RefSeq) database at
33	NCBI: current status, taxonomic expansion, and functional annotation. Nucleic Acids Res
34	44(D1):D733-45.
35	Perrault I, Saunier S, Hanein S, Filhol E, Bizet AA, Collins F, Salih MA, Gerber S, Delphin
36	N, Bigot K and others. 2012. Mainzer-Saldino syndrome is a ciliopathy caused by IFT140
37	mutations. Am J Hum Genet 90(5):864-70.
38	Redin C Le Gras S Mhamdi O Geoffroy V Stoetzel C Vincent MC Chiurazzi P Lacombe
39	D Ouertani I Petit F and others 2012 Targeted high-throughput sequencing for diagnosis of
40	genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and Alstrom
41	syndromes I Med Genet 49(8):502-12
42	Reese MG Feckman FH Kuln D Haussler D 1997 Improved splice site detection in Genie
43	L Comput Biol 4(2):211-22
44 45	Bice D. Longdon I. Bleachy A. 2000 EMBOSS: the European Malagular Dialogy Open
45	Kice P, Longuell I, Bleasby A. 2000. EMBOSS. the European Molecular Biology Open Software Swite Tranda Canat 16(6):276.7
40	Soliware Suile. Hends Genet 10(0).270-7.
47	Schaefer E, Stoetzel C, Scheidecker S, Geoffroy V, Prasad MK, Redin C, Missotte I,
40	Lacombe D, Mandel JL, Muller J and others. 2016. Identification of a novel mutation
50	confirms the implication of IFTT/2 (BBS20) in Bardet-Biedl syndrome. J Hum Genet
51	61(5):44/-50.
52	Scheidecker S, Etard C, Pierce NW, Geoffroy V, Schaefer E, Muller J, Chennen K, Flori E,
53	Pelletier V, Poch O and others. 2014. Exome sequencing of Bardet-Biedl syndrome patient
54	identifies a null mutation in the BBSome subunit BBIP1 (BBS18). J Med Genet 51(2):132-6.
55	Schmidts M, Frank V, Eisenberger T, Al Turki S, Bizet AA, Antony D, Rix S, Decker C,
56	Bachmann N, Bald M and others. 2013. Combined NGS approaches identify mutations in the
57	
58	
59	20
60	John Wiley & Sons, Inc. 20

intraflagellar transport gene IFT140 in skeletal ciliopathies with early progressive kidney Disease. Hum Mutat 34(5):714-24.

Schneider CA, Rasband WS, Eliceiri KW. 2012. NIH Image to ImageJ: 25 years of image analysis. Nat Methods 9(7):671-5.

Shapiro MB, Senapathy P. 1987. RNA splice junctions of different classes of eukaryotes: sequence statistics and functional implications in gene expression. Nucleic Acids Res 15(17):7155-74.

Sherry ST, Ward MH, Kholodov M, Baker J, Phan L, Smigielski EM, Sirotkin K. 2001. dbSNP: the NCBI database of genetic variation. Nucleic Acids Res 29(1):308-11.

Stankiewicz P, Sen P, Bhatt SS, Storer M, Xia Z, Bejjani BA, Ou Z, Wiszniewska J, Driscoll DJ, Maisenbacher MK and others. 2009. Genomic and genic deletions of the FOX gene cluster on 16q24.1 and inactivating mutations of FOXF1 cause alveolar capillary dysplasia and other malformations. Am J Hum Genet 84(6):780-91.

Stoetzel C, Bar S, De Craene JO, Scheidecker S, Etard C, Chicher J, Reck JR, Perrault I, Geoffroy V, Chennen K and others. 2016. A mutation in VPS15 (PIK3R4) causes a ciliopathy and affects IFT20 release from the cis-Golgi. Nat Commun 7:13586.

Tan R, Wang Y, Kleinstein SE, Liu Y, Zhu X, Guo H, Jiang Q, Allen AS, Zhu M. 2014. An evaluation of copy number variation detection tools from whole-exome sequencing data. Hum Mutat 35(7):899-907.

Taylor JC, Martin HC, Lise S, Broxholme J, Cazier JB, Rimmer A, Kanapin A, Lunter G, Fiddy S, Allan C and others. 2015. Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nat Genet 47(7):717-726.

Thorvaldsdottir H, Robinson JT, Mesirov JP. 2013. Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. Brief Bioinform 14(2):178-92. Tyner C, Barber GP, Casper J, Clawson H, Diekhans M, Eisenhart C, Fischer CM, Gibson D, Gonzalez JN, Guruvadoo L and others. 2017. The UCSC Genome Browser database: 2017 update. Nucleic Acids Res 45(D1):D626-D634.

Yeo G, Burge CB. 2004. Maximum entropy modeling of short sequence motifs with applications to RNA splicing signals. J Comput Biol 11(2-3):377-94.

Zhu B, Zhu X, Wang L, Liang Y, Feng Q, Pan J. 2017. Functional exploration of the IFT-A complex in intraflagellar transport and ciliogenesis. PLoS Genet 13(2):e1006627.

Figure legends

Figure 1. Identification of 2 mutations in *IFT140* for a family explored by WGS.

(A) Pedigree of family A, which has 2 affected individuals. Segregation analysis of the 2 mutations noted M1: 2577+25G>A and M2: 3454-488_4182+2588dup. Example of Sanger sequencing profiles for individual II.1. The breakpoint junction between exon 30 and intron 26 are shown.

(B) M1 analysis. Sanger sequencing was performed on RNA extracted from fibroblasts of individual II.1 and a healthy unrelated control amplified between exon 18 and exon 22.

(C) M2 analysis. Quantitative real-time PCR was performed on DNA from exon 30 in all individuals from family A and one unrelated control. DNA quantity from exon 30 of *IFT140* was compared to 2 reference genes (*HBB* and *HMBS*) using the absolute quantification method.

(D) M2 RNA expression in patient's II.1 skin fibroblasts is demonstrated by PCR amplification (*PRMT9* as a control) and by Sanger sequencing.

(E) IFT140 expression in skin fibroblasts was revealed by western blot using anti-IFT140 antibody.

Figure 2. Patients' fibroblasts have reduced number of ciliated cells and mislocalized IFT140

(A) Number of ciliated cells were counted on serum deprived (24H) control and patient's skin fibroblasts fixed for immunofluorescence and stained with anti-IFT140 (red) and anti-acetylated tubulin (green) antibodies. Nuclei were colored in blue (Dapi).

(B) Based on 15 fields in three independent experiments (100-200 cells per experiment), mean percentages of ciliated cells are shown in a histogram together with corresponding standard deviation and p-values (n.s.: non-significant, *: p < 0.0001).

(C) IFT140 localisation was assessed on serum deprived (24H) control and patient's skin fibroblasts fixed for immunofluorescence and stained with anti-IFT140 (red) and anti-acetylated tubulin (green) antibodies. Nuclei were colored in blue (Dapi).

(D) Primary cilia containing IFT140 were counted based on three independent experiments (60-200 ciliated cells per experiment), mean percentages are shown in a histogram together with corresponding standard deviation and p-values (n.s.: non-significant, *: p<0,0005).

Figure 3. Mutations in *IFT140*

(A) Pedigrees of 7 additional families with affected individuals carrying the tandem duplication.

(B) Pedigrees of additional families with *IFT140* pathogenic variants.

Figure 4. Schematic representation of *IFT140* and of the tandem duplication

(A) *IFT140* extends over 105.6 kb in chromosomal region 16p13.3 and contains 31 exons encoding a 1462 amino acids protein containing two repeated domains, 5 WD repeats and 9 tetratricopeptide (TPR) repeats. Previously reported mutations in *IFT140* are represented using a black "asterisk" whereas novel mutations are colored in red. In particular, the two mutations from family A are also shown in introns 20 (c.2577+25G>A) and 30 (c.3454-490_4182+2588dup). The tandem duplication (black horizontal bar) spans from exon 27 to 30 (6.7 kb). The breakpoints are located within *Alu* elements (*AluJb* and *AluJr*).

 (B) Breakpoint junction sequence of affected individuals with tandem duplication. Breakpoint junction sequence is aligned to the proximal and distal genomic references, respectively *AluJr* (chr16:1565570-1565697) and *AluJb* (chr16:1571239-1571365), and color-matched. Microhomology at the breakpoint is indicated in red. The aligned genomic region is 128 bp long showing 81% of sequence identity.

(C) Proposed rearrangement of Alu-Alu mediated duplication in affected individuals.

to pee periev



Α

С

30]/[Reference]



59

60

c.[M1];[=] в TGGGGTGA GTGGGAAGGCA Control GTG AGG GGC GCC CGC CAT GGG Arg Gly Arg Arg His 18 19 20 21 22 18 19 20 21 22 TGAGCCCAGGAGTTCA Mamman A-II.1 Control D 1.8 1,6 CATGAGGG 1,4 1,3 1 0,6 Ę V-II.2 Е A-II.1 A-II.2 A-I.2 A-I.1 250 - IFT140 (165 kDa) 150

Figure 1. Identification of 2 mutations in IFT140 for a family explored by WGS.

(A) Pedigree of family A, which has 2 affected individuals. Segregation analysis of the 2 mutations noted M1: 2577+25G>A and M2: 3454-488_4182+2588dup. Example of Sanger sequencing profiles for individual II.1. The breakpoint junction between exon 30 and intron 26 are shown.

(B) M1 analysis. Sanger sequencing was performed on RNA extracted from fibroblasts of individual II.1 and a healthy unrelated control amplified between exon 18 and exon 22.

(C) M2 analysis. Quantitative real-time PCR was performed on DNA from exon 30 in all individuals from family A and one unrelated control. DNA quantity from exon 30 of IFT140 was compared to 2 reference genes (HBB and HMBS) using the absolute quantification method.

(D) M2 RNA expression in patient's II.1 skin fibroblasts is demonstrated by PCR amplification (PRMT9 as a control) and by Sanger sequencing.

(E) IFT140 expression in skin fibroblasts was revealed by western blot using anti-IFT140 antibody.

297x166mm (300 x 300 DPI)





Figure 2. Patients' fibroblasts have reduced number of ciliated cells and mislocalized IFT140 (A) Number of ciliated cells were counted on serum deprived (24H) control and patient's skin fibroblasts fixed for immunofluorescence and stained with anti-IFT140 (red) and anti-acetylated tubulin (green) antibodies. Nuclei were colored in blue (Dapi).

(B) Based on 15 fields in three independent experiments (100-200 cells per experiment), mean percentages of ciliated cells are shown in a histogram together with corresponding standard deviation and p-values (n.s.: non-significant, *: p< 0,0001).

(C) IFT140 localisation was assessed on serum deprived (24H) control and patient's skin fibroblasts fixed for immunofluorescence and stained with anti-IFT140 (red) and anti-acetylated tubulin (green) antibodies. Nuclei were colored in blue (Dapi).

(D) Primary cilia containing IFT140 were counted based on three independent experiments (60-200 ciliated cells per experiment), mean percentages are shown in a histogram together with corresponding standard deviation and p-values (n.s.: non-significant, *: p<0,0005).

220x141mm (300 x 300 DPI)

c.[634G>A];[3454-488_4182+2588dup]

Family G

Family K

1 (

c.[?];[=]

c.[1319T>C];[=]

c.[3454-488 4182+2588dup];[=]

c.[1-7_147 +?del];[=]

?del];[1319T>C

c.[3454-488_4182+2588dup];[3454-488_4182+2588d

Family H

c.[2177C>T];[3454-488_4182+2588dup]





Figure 4. Schematic representation of IFT140 and of the tandem duplication
(A) IFT140 extends over 105.6 kb in chromosomal region 16p13.3 and contains 31 exons encoding a 1462 amino acids protein containing two repeated domains, 5 WD repeats and 9 tetratricopeptide (TPR) repeats. Previously reported mutations in IFT140 are represented using a black "asterisk" whereas novel mutations are colored in red. In particular, the two mutations from family A are also shown in introns 20
(c.2577+25G>A) and 30 (c.3454-490_4182+2588dup). The tandem duplication (black horizontal bar) spans from exon 27 to 30 (6.7 kb). The breakpoints are located within Alu elements (AluJb and AluJr).
(B) Breakpoint junction sequence of affected individuals with tandem duplication. Breakpoint junction sequence is aligned to the proximal and distal genomic references, respectively AluJr (chr16:1565570-1565697) and AluJb (chr16:1571239-1571365), and color-matched. Microhomology at the breakpoint is indicated in red. The aligned genomic region is 128 bp long showing 81% of sequence identity. (C) Proposed rearrangement of Alu-Alu mediated duplication in affected individuals.

232x153mm (300 x 300 DPI)

SUPPLEMENTARY METHODS

Whole Exome Sequencing

Whole Exome Sequencing (WES) was performed for the two affected siblings from family A (A-II.1 and A-II.2) by the IGBMC (Institute of Genetics and Molecular and Cellular Biology) Microarray and Sequencing platform. Exons of DNA samples were captured with in-solution enrichment methodology (Agilent SureSelect All Exon XT2 50 Mb Kit) and sequenced with an Illumina HiSeq 2500 instrument as paired-end 100 bp reads, in order to reach an average sequencing depth of 80x for each sample. The sequence reads were aligned to the reference sequence of the human genome (GRCh37) using the Burrows-Wheeler Aligner (BWA V7.12) (Li and Durbin, 2010). The HaplotypeCaller module of the Genome Analysis ToolKit (GATK, v.3.4.46) (DePristo, et al., 2011) was used for calling both SNV and indel.

Targeted Exome Sequencing

Leber panel

Targeted exome sequencing of 14 full genes related to Leber congenital amaurosis was performed for 126 patients (including family B, C, D, E and I). Illumina compatible precapture barcoded genomic DNA libraries were constructed according to the manufacturer's sample preparation protocol (Ovation Ultralow, Nugen Technologies). Exons of DNA samples were captured with in-solution enrichment methodology (Agilent SureSelect custom panel) and sequenced with an Illumina HiSeq2500 (Paired-End sequencing 2x125 bases, Rapid Mode, 80 libraries per lane). Bioinformatics analysis was performed as described in Gerber *et al (Gerber, et al., 2016)* using an in house pipeline (POLYWEB).

RP panel

Targeted exome sequencing of 267 genes related to retinitis pigmentosa was performed for 117 patients (including family J). Exons of DNA samples were captured with in-solution enrichment methodology (Agilent QXT SureSelect custom panel) and sequenced with an Illumina NextSeq 550 instrument (Paired-End sequencing 2x150 bases, 48 libraries per lane). SNVs and indels were called with the Genome Analysis Toolkit v.3.4.46 thanks to our in house pipeline (STARK) and following the GATK best practice.

Ciliome panel

Targeted exome sequencing of 1221 ciliary candidate genes including genes related to ciliopathies like Jeune asphyxiating thoracic dystrophy, Mainzer-Saldino syndrome, Bardet-Biedl syndrome, Joubert syndrome and Senior Loken was performed for 364 patients (including families C, D, I and K). Sequencing was conducted using a custom SureSelect capture kit (Agilent Technologies) targeting 4.5 Mb of 20,168 exons, and performed on SOLiD5500XL (Life Technologies) or HiSeq (Illumina). Bioinformatics analysis was performed as described in Grampa *et al* (Grampa, et al., 2016) using an in house pipeline (POLYWEB). The mean depth of coverage obtained was greater than 165x, with \geq 89% of the bases covered at least 15x. In order to evaluate duplication and large deletion events, for each individual the relative read count for each targeted region was determined as the ratio of the read count for that region divided by the total absolute read counts of all targeted regions of the design. The ratio of the relative read count of a region in a given individual over the average relative read counts in other individuals of the run resulted in the estimated copy number for that region in that individual.

Missense pathogenicity analysis

For the 2 novel missenses identified in this study, conservation has been assessed using the multiple sequence alignment available from the eggnog database (KOG3717, (Huerta-Cepas, et al., 2016)) and they were analyzed thanks to the PolyPhen-2, SIFT and MutationTaster softwares (Adzhubei, et al., 2010; Kumar, et al., 2009; Schwarz, et al., 2010)

to per perie

Disease	Allele 1	Location	Allelic count	Allele 2	Location	Allelic count	Family
Perrault et	<i>t al</i> 2012 (Perrault, et al., 2012) and this study						1
MSS	c.2399+1G>T (p.?)	Intron 19	5/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
MSS	c.932A>G (p.Tyr311Cys)	Exon 9	1/120	c.857_860del (p.Ile286Lysfs*6)	Exon 8	1/120	
MSS	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
MSS	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
MSS	c.634G>A (p.Gly212Arg)	Exon 7	7/120	c.3916dupG (p.Ala1306Glyfs*56)	Exon 29	1/120	
MSS	c.699T>G (p.Ile233Met)	Exon 7	2/120	c.699T>G (p.Ile233Met)	Exon 7	2/120	
JATD	c.2399+1G>T (p.?)	Intron 19	5/120	c.634G>A (p.Gly212Arg)	Exon 7	7/120	
MSS	c.1565G>A (p.Gly522Glu)	Exon 14	4/120	c.3454-488_4182+2588dup p.(Tyr1152_Thr1394dup)	Introns 26 30	10/120	Family B
JATD/MSS	c.874G>A (p.Val292Met)	Exon 8	4/120	c.3454-1005_4040+737delinsCCC, (p.Tyr1152Aspfs*14)	Exons 27-30	1/120	Family I
MSS	c.1727G>A (p.Arg576Gln)	Exon 15	1/120	2 nd allele not found			
MSS	c.489C>T (p.Gly163Gly), and c.488_491del (p.Glu164Thrfs*10) from creation of additional donor splice site	Exon 5	1/120	2 nd allele not found			
Schmidts a	<i>et al</i> 2013 (Schmidts, et al., 2013)			·			
JATD	c.1380delC (p.Asn460Lysfs*28)	Exon 12	1/120	c.874G>A (p.Val292Met)	Exon 8	4/120	
JATD	c.1565G>A (p.Gly522Glu)	Exon 14	4/120	c.874G>A (p.Val292Met)	Exon 8	4/120	
JATD	c.1565G>A (p.Gly522Glu)	Exon 14	4/120	c.874G>A (p.Val292Met)	Exon 8	4/120	
JATD	c.454C>T (p.Leu152Phe)	Exon 5	3/120	c.454C>T (p.Leu152Phe)	Exon 5	3/120	
JATD	c.2278C>T (p.Arg759*)	Exon 19	2/120	2 nd allele not found			
MSS	c.2399+1G>T (p.?)	Intron 19	5/120	c.4078T>C (p.Cys1360Arg)	Exon 30	1/120	
MSS	c.418G>A (p.Gly140Arg) and c.800A>G (p.Glu267Gly)	Exons 5/7	1/120	c.490G>T (p.Glu164*)	Exon 6	1/120	
JATD	c.2399+1G>T (p.?)	Intron 19	5/120	2 nd allele not found			
JATD	c.4058 G>C (p.Pro1353Arg)	Exon 30	1/120	2 nd allele not found			
JATD	c.2360 T>C (p.Asp787Gly)	Exon 19	1/120	2 nd allele not found			
JATD	c.1541 A>T (p.Leu514His)	Exon 14	1/120	2 nd allele not found			
JATD	c.329G>A (p.Arg110His)	Exon 4	1/120	2 nd allele not found			
JATD	c.481C>A (p.Pro161Thr)	Exon 5	2/120	2 nd allele not found			

JATD	c.481C>A (p.Pro161Thr)	Exon 5	2/120	2 nd allele not found			
Khan <i>et a</i>	d 2014 (Khan, et al., 2014)						
MSS	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
MSS	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
Bifari <i>et a</i>	ul 2015 (Bifari, et al., 2016)						
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	c.1990G>A (p.Glu664Lys)	Exon 17	31/120	
EORD	c.1541_1542delinsAA (p.Leu514Gln)	Exon 14	2/120	c.1541_1542delinsAA (p.Leu514Gln)	Exon 14	2/120	
Xu et al 2	015 (Xu, et al., 2015)						
RP	c.4196T>C (p.Leu1399Pro)	Exon 31	31/120	c.1898_1901delATAA (p.Asn633Serfs*10)	Exon 17	1/120	
RP	c.3826G>A (p.Gly1276Arg)	Exon 28	1/120	c.1989C>G (p.Cys663Trp)	Exon 17	1/120	
RP	c.650_651delTG (p.Val217Glyfs*2)	Exon 7	1/120	c.212C>T (p.Pro71Leu)	Exon 4	1/120	
RP	c.2611C>T (p.Arg871Cys)	Exon 21	1/120	c.1377G>A (p.Trp459*)	Exon 12	1/120	
RP	c.1252G>C (p.Ala418Pro)	Exon 11	1/120	c.2921C>T (p.Ala974Val) presumed in trans	Exon 23	1/120	
LCA	c.1451C>T (p.Thr484Met)	Exon 13	3/120	c.985T>C (p.Cys329Arg)	Exon 9	1/120	
LCA	c.2368G>A (p.Glu790Lys)	Exon 19	1/120	c.1655_1656delAG (p.Glu522Glyfs*6)	Exon 14	1/120	
Beheshtia	n <i>et al</i> 2015 (Beheshtian, et al., 2015)						
RP	c.3827G>A (p.Gly1276Glu)	Exon 28	2/120	c.3827G>A (p.Gly1276Glu)	Exon 28	2/120	
Weisschu	h <i>et al</i> 2016 (Weisschuh, et al., 2016)						
RP	c.472C>T (p.Arg158Trp)	Exon 5	1/120	c.1565G>A (p.Gly522Glu)	Exon 14	4/120	

RP	c.1451C>T (p.Thr484Met)	Exon 13	3/120	c.1451C>T (p.Thr484Met)	Exon 13	3/120	
RP	c.2399+1G>T (p.?)	Intron 19	5/120	c.2815T>C (p.Ser939Pro)	Exon 22	1/120	
RP	c.998G>A (p.Cys333Tyr)	Exon 9	4/120	c.998G>A (p.Cys333Tyr)	Exon 9	4/120	
RP	c.998G>A (p.Cys333Tyr)	Exon 9	4/120	c.998G>A (p.Cys333Tyr)	Exon 9	4/120	
RP	c.1021G>A (p.Ala341Thr)	Exon 10	1/120	c.1422_23insAA (p.Arg475Asnfs*14)	Exon 12	1/120	
Pena-Padi	lla <i>et al</i> 2016 (Pena-Padilla, et al., 2016)						
OTCS	c.3141+1G>T (p.?)	Exon 24	2/120	c11_6del (p.?)	Exon 3	1/120	
Bayat <i>et al</i>	2017 (Bayat, et al., 2017)						
SB	c.634G>A (p.Gly212Arg)	Exon 7	7/120	c.2278C>T (p.Arg759*)	Exon 19	2/120	
Helm <i>et al</i>	2017 (Helm, et al., 2017)						
MSS	c.634G>A (p.Gly212Arg)	Exon 7	7/120	c.634G>A (p.Gly212Arg)	Exon 7	7/120	
This study							
MSS	c.2577+25G>A (p.Leu859_Glu860insValArgGlyAlaArgHisGly)	Intron 20	1/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family A
MSS	c.634G>A (p.Gly212Arg)	Exon 7	7/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family C
MSS	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family D
JATD/MSS	c.3141+1G>T (p.?)	Exon 24	2/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family E
MSS	c.454C>T (p.Leu152Phe)	Exon 5	3/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family F
MSS	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family G
MSS/SB	c.2177C>T (p.Pro726Leu)	Exon 18	1/120	c.3454-488_4182+2588dup (p.Tyr1152_Thr1394dup)	Introns 26-30	10/120	Family H
RP	c.634G>A (p.Gly212Arg)	Exon 7	7/120	c.4236_4239dup (p.Tyr1414Leufs*48)	Exon 31	1/120	Family J
MSS (mild)	c.1-?_147+?del, start loss	Exon 3	1/120	c.1319T>C (p.Leu440Pro)	Exon 11	1/120	Family K

Table S1. Previously reported and novel mutations in *IFT140*.

Mutations are reported only once per family. Those reported in this study are in bold and those described for the first time are also in italic. Mutation allelic counts are reported for only one affected individual per family. It is to notice that some studies (i.e. Bifari *et al*) have analyzed consanguineous families from the same region increasing the global allelic count. EORD: early-onset retinal dystrophy; JATD: Jeune asphyxiating thoracic dystrophy; MSS: Mainzer-Saldino Syndrome; OCTS: Opitz trigonocephaly C syndrome; RP: retinitis pigmentosa; SB: Sensenbrenner syndrome.

3												
4												
5 Individual	A-II.1	A-II.2	B-II.1	C-II.2	D-II.1	E	F	G	Н	I-II.1	J-II.1	K-II.2
J Birth	10/01/1998	31/08/1999 Engine Italy	17/03/1978 Sarbia Vicenslavia	26/08/1976	27/12/1968	20/09/2010	11/02/2007	21/05/1993		16/04/1986	05/06/1993	14/02/2008
6 5.	M	M	E Serbia, i ugosiavia	Flance	Flance	Caucasian	Ausura	Ausula	E	м	м	E
Suspected diagnostic	MSS	MSS	MSS	MSS	MSS	1' Jeune/MSS	MSS	MSS	1 MSS/Sensenbrenner	Ivi Jeune/MSS	Ivi Isolated RP	MSS (mild)
7 Onset	14 yo	15 vo	3 v0	6 vo	45 vo	Neonatal	4 vo	1 st vo	Neonatal	Jeane/10155	130iated Iti	7 vo
Short stature (SD)	+ (-3 SD)	+ (-2 SD)	+ (< - 5 SD)	+ (- 3 SD)	+ (< - 5 SD)	+ disproportionate (-3 SD)	(-1 SD)	(+1,5 SD)			(-1 SD)	.).
8 Skeletal features 9 10 11	Broad long bones	Hip anomalies (narrow pelvis with narrow iliac wings, lack of coverage of superior femoral epiphyses)	Hip anomalies Metaphyseal defect (forearms) Micromelia Scoliosis	Metaphyseal defect (long bones)	Short and broad bones Bilateral ulnar valgus Marked curved radius Hip anomalies (large pelvis, short femoral heads) Varus of the feet	Hypoplastic thorax. Short, slightly broader ribs Hip anomalies (hip dysplasia, broad epiphyses, steep acetabular roofs) Genu varum Hyperlordosis Brachymelia No premature craniosynostosis	Moderate hips' anomalies	No	Craniostenosis Hip anomalies not known	Thoracic dystrophy		
12 ^{Digit anomalies} 13 14 15	Brachymetacarpia (1, 3, 4 digits) Brachymetatarsia	Short hands Brachydactyly and brachymetacarpia (1, 4 and 5 digits) brachymetatarsia	Short hands Cone-shaped phalangeal epiphyses (hands)	Cone-shaped phalangeal epiphyses (hands)	Brachytelephalangy Brachymesophalangy with clinodactyly of the 5 th digits (feet and hands) Enlarged metaphyses (feet) No cone-shaped epiphyses due to the age of the patient (52 yo)	Short hands Brachyphalangy Cone-shaped phalangeal epiphyses (hands) Pes planovalgus Incomplete syndactyly of digits 3-4 (toes) Brachydactyly of 3rd toe Delayed bone age	Short hands Brachymesophalangy Cone-shaped phalangeal epiphyses (feet, hands)	Short hands Brachymesophalangy Cone-shaped phalangeal epiphyses (feet, hands)	Short hands Brachymesophalangy Cone-shaped phalangeal epiphyses No polydactyly	Cone- shaped phalangeal epiphyses		Cone-shaped phalangeal epiphyses (second phalanges of the 2, 3, 4 digits)
16 17 18	Hemeralopia (10 yo) VA: 9/10RLE	Hemeralopia (7 yo) Divergent strabismus VA: 7/10RLE RP Altered ERG	Onset (3 mo) Nystagmus Night blindness VA: 2/10RLE Maculopathy Altered ERG	VA: 4/10RLE	Divergent strabismus (2 yo) Hemeralopia (10 yo) RP (16 yo) VA: 2.4/10 RE 1/20 LE Advanced RP VF: very reduced on right side	Congenital pendular nystagmus Strahismus Hyperopia Astigmatism Severe visual reduction	Myopia RP (4 yo)	Myopia RP (1 yo)	Nystagmus Strabismus Refraction +2 Retinal dystrophy not known Papillae edges not well differentiated	ERG: no signal left	RP slow evolution	No eye pursuit (1 mo) Bilateral divergent strabismus (5 mo) Nystagmus (6 mo) Early onset advanced RP (3 yo) ERG: severe and primary damage of cones and rods
20 21 22 23 24	Moderate renal failure Microalbuminuria Renal US normal	Moderate renal failure Microalbuminuria Renal US normal Arterial hypertension	End stage renal failure (5 yo) Dialysis Transplant Nephronophtissis with polyria polydipsia (3 yo) Arterial hypertension Renal US (Several cortical cysts (10 yo), echogenicity, kidney's size: R76mm L60mm)	End stage renal failure (7 yo) Dialysis Transplant (7 yo) Neptronophitisis with polyria polydipsia (6 yo) Arterial hypertension (24/12) Renal US (no cysts, echogenicity, kidney's size: R55mm L59mm) Obsolescent/hyalinized glomerulus tubulo-intersticial nephritis important arterial damage	End stage renal failure Dialysis (10 yo) Transplantation (16 yo)	End stage renal failure (1 5 yo) Peritoneal dialysis (2 yo) Transplant (2 5 yo) Hydronephrosis (4 mo) Renal US (small hyperechoic kidney, right kidney with 3 chambered cysts) Artenial hypertension Renal anemia	End stage renal failure (4.7 yo) Renal US (Small kidneys (<3th centile, increased echogenicity, reduced cortico-medullar differentiation, no cysts) Proteinuria, microhematuria	End stage renal failure (23 yo) Renal US (cysts, low- normal size, increased echogenicity, loss of cortico-medullar differentiation (11 yo) Glomerat and tubular proteinuria	End stage renal failure Dialysis (3.8 yo) Transplant CKD stage V Renal US (small cysts, small kidney) Glomerular and tubular proteinuria No hematuria			Renal function normal Renal ultrasound normal
Hepatic involvement	No	No	Portal space fibrosis	· · · · · · · · · · · · · · · · · · ·	No	Slightly hyperechoic liver	Yes	No	Normal liver function tests			
25 Barrier Charles	Ne	Ne	Hemosiderosis		Ne	Slightly enlarged			Hepatomegaly	Van (aunti)		
26 Neurological features 27 28	No	No	No		INO	Asphyxia at birth Slight Developmental delay (head control 4 mo, sitting 8 mo, walking 24 mo, first words 18 mo, slow speech development) No ataxia, no seizures No autistic features	No	Hypotonia at birth Developmental delay	Developmental delay No cerebellar ataxia, but oculomotor apraxia Intracranial hypertension No seizures	res (cysuc)		
29 Cerebral anomalies	No	No				Generalized brain atrophy (CT scan 4 mo)	No		No		No	No
30 Dysmorphic features 31	Yes (high hairline, frontal bossing, ptosis, long philtrum, prominent chin)	Yes (high forehead, frontal bossing, long philtrum, prominent chin)			No	Yes (high hairline, frontal bossing, hypertelorism, broad nasal ridge, deep-set eyes, anteverted nares, long philtrum, everted lower lip, prominent chin, deep- set ears)	No	No	Yes			
3P Oral anomalies	No	No	No		No	No	No	No	No			
33 24	Asthma					Truncal adiposity, breast tissue Splenomegaly Obstructive bronchitis Recurrent infections	Recurrent respiratory Infections	Late onset obesity	Premature birth 28 weeks of gestational age		Overweight when younger (9 yo)	Joint luxations with spontaneous subluxations
35 36	Table S2	Clinical Feat	ures of MSS-a	ffected individua	ls harboring co	ompound-heterozygo	ous mutatior	is in <i>IFT140</i> .	CKD: Cystic F	Kidney I	Disease,	

Table S2 Clinical Features of MSS-affected individuals harboring compound-heterozygous mutations in *IFT140*. CKD: Cystic Kidney Disease,
CT: Computed Tomography, ERG: Electroretinogram, F: Female, L: Left, LE: Left Eye, M: Male, mo: month old, MSS: Mainzer-Saldino
Syndrome, R: Right, RE: Right Eye, RLE: Right and left Eye, RP: Retinitis Pigmentosa, SD: standard deviation, US: Ultrasound, VA: Visual
acuity, VF: visual field, yo: years old.

1	
2	
3	
4	
5	
6	
7	
8	
9	
10	
11	
12	
13	
14	
15	
10	
10	
10	
20	
21	
22	
23	
24	
25	
26	
27	
28	
29	
30	
31	
32	
33	
34	
35	
36	
37	
38	
39 40	
40 1	
41 ⊿ว	
42 12	
43 44	
45 45	
чJ	

Application	Gene	RefSeq	Name	Forward (5'-3')	Reverse (5'-3')
DNA c.2577+25G>A	IFT140	NM_014714.3	IFT140-ex20	GATGACTACTTGCCCCAGGA	CACACCTGACAGGCACACAC
DNA breakpoint TD			IFT140-intron30F_intron26R	CCGGAGTAGCTGGGATTACA	CTGTCTAGCTGGGGTGGGTA
DNA c.1565G>A			IFT140-ex14	TGAGCGAGTGAATGAGTGGA	CGGTGTTAAAATGGGCACCT
cDNA c.2577+25G>A			IFT140-RT-PCR-ex18-19F_ex22R	ATCAAAAGTGAGGCCGTCTG	GCGTGTCCGACTTCTCGTA
cDNA TD			IFT140-RT-PCR-ex30F_ex28R	AAGCAGTGTGAGCTGCTCCT	GGGCACAAGCGTCATAAAAG
cDNA control	PRMT9	NM_001304458	PRMT9-RT-PCR-ex3F_ex6R	GTGGAACGCTGGCACTTTAT	CAATTAGCACTTTCACCTTTGG
qPCR	IFT140	NM_014714.3	IFT140-QPCR-ex30F_ex30R	CATAGAAAGTTTCCTCCTGCCAT	TCTGGTTCCTCCAGGAGCA
qPCR	HMBS	NM_000190	HMBS-QPCR-	CACAGCACTCCCACTGACAAC	GAGGGAGGCGATAGTAGGACC
qPCR	HBB	NM_000518	BGLOB-QPCR-ex1F_ex1R	ACACAACTGTGTTCACTAGC	CAACTTCATCCACGTTCACC
Duplex PCR	IFT140	NM_014714.3	IFT140-intron30F_intron26R	CCCAAAGCGTCATTTCTGGTGTTC	CCCAGGGTCAGGTGATCTTCCTA
	BBS12	NM_152618	BBS12-ex2F_ex2R	AGTTCTCATTGAGGGTGACCT	GCCAGAGATGAAGCCAGCCA

Table S3 List and characteristics of primers used in this study. TD: Tandem duplication of exon 27-30

2
2
5
4
5
6
7
, 0
ð
9
10
11
12
12
13
14
15
16
17
17
18
19
20
20
21
22
23
24
25
25
26
27
28
20
29
30
31
32
22
24
34
35
36
37
20
20
39
40
41
12
42
43
44
45
46
-+U /-7
4/
48
49
50
50 E 1
21
52
E 2

Name primary antibodies	Applications	Producer	Conditions
Mouse anti-Acetylated α-Tubulin	WB, IF	Abcam #ab24610	IF: 1/200
Rabbit anti-IFT140	WB, IF	Proteintech #17460-1-AP	IF:1/100 WB: 1/1000
Mouse anti-β-Tubulin	WB	Euromedex # TUB-2A2	1/2500
Name secondary antibodies			
Donkey anti-Mouse IgG (H+L), FITC conjugate	IF	Thermo Scientific #A16012	1/500
Goat anti-Rabbit IgG (H+L) Alexa Fluor 568 conjugate	IF	Thermo Scientific #A-11011	1/500
Chicken anti-Rabbbit IgG-HRP	WB	Santa Cruz #sc-2955	1/5000
Goat anti mouse IgG-HRP	WB	Santa Cruz #sc-2060	1/5000

Table S4. List of antibodies used in this study. IF: immunofluorescence, WB Western Blot.

K PER PENE



Figure S1. Duplex PCR profiles on a LabChip GX (Perkin Elmer) for a rapid detection of the *IFT140* tandem duplication (see **Table S3** for primers details). Normal samples harbor a single band (*BBS12*) whereas homozygous or heterozygous carriers for the tandem duplication show an additional band specific for the duplication breakpoint.

2	
3	
4	
5	
6	
7	
8	
0 0	
9 10	
10	
11	
12	
13	
14	
15	
16	
17	
10	
10	
19	
20	
21	
22	
23	
24	
25	
26	
20	
27	
28	
29	
30	
31	
32	
33	
34	
35	
36	
20	
3/	
38	
39	
40	
41	
42	
43	
44	
45	
75 76	
40	
4/	
48	
49	
50	
51	
52	
53	
54	
54	
22	
56	
57	
58	
59	

1

Patient	A-l	I.1	A-l	1.2
Type of sequence variant	SNV	indel	SNV	indel
Total number of variants	49489	2168	49418	2133
After exclusion of variants with an allele frequency $>1\%$ (extracted from dbSNP, the Exome Variant Server and the ExAC database or our internal exome database)	6528	1038	6517	1017
After exclusion of variants found at the homozygous state and more than once at the heterozygous state in 90 control exomes	1159	90	1029	72
After exclusion of 5'UTR, 3'UTR, downstream, upstream and intron locations without local splice effect prediction (from the "localSpliceEffect" field of Alamut-Batch)	517	20	458	12
After exclusion of synonymous variants without local splice effect prediction (from the "localSpliceEffect" field of Alamut-Batch)	417	20	373	12
After selection of variants consistent with recessive transmission (compound heterozygous, homozygous variants).	2 co	mpound h PLE	eterozygo KHA8	us in

Table S5 Summary of the whole exome sequencing results from family A

WES data processing and variant calling revealed respectively 51,657 and 51,551 SNV and indels in individual A-II.1 and A-II.2. Variant filtering using stringent criteria reduced the number of genetics variants to respectively 437 and 385 variants per proband. To identify variants consistent with autosomal recessive inheritance, we kept only compound shared heterozygous or homozygous variants, reducing the number of variants to 2 heterozygous variants in the *PLEKHA8* gene: NM_001197026.1:c.1517_1518insAT (p.Leu507Phefs*27) and NM_001197026.1:c.1522del (p.Tyr508Metfs*25). Both variants being always supported on the same reads (as shown in the IGV view below), they are in *cis* and are probably an artifact due to a bad reads alignment. So, no candidate gene was found by the WES analyses.

IGV	- T- KIK B * MARCO MARCO MARCO AND	
<u>File</u> Genomes <u>View</u> Tra-	ic <u>k</u> s Regions Tools GenomeSpace Help	
Human (b37)	▼ 7 → 7:30,118,327-30,118,419 Go 🚔 ◀ ▶ 🆓 🗖 🗶 🖵 🖃	IIIII <mark>I</mark> I I
	p221 p212 p153 p143 p141 p122 p113 q1122 q2131 q212 q223 q313 q3131 q3131 q323 q3	5 q36.2
	←93 bp90 118 200 bp 20 118	bp 30 11
	¥ = 1	ŕ
	II =	
	I H	=
VOLF_HDS-02.aligned.sorted.de .realign.recal.bam		
		-
Sequence →	L C PLW C C A S A C C A L C A C C L C A C C C C C C C C	G Q H G S
Gene		
6 tracks loaded 7:30	118 416 44	3M of 590M



Figure S2. Next generation sequencing data from patient A-II.1 displayed around *IFT140*, in particular from exon 27 to exon 30 highlighting the tandem duplication.

(A) Comparison between whole genome sequencing (WGS) and whole exome sequencing (WES) data from patient A-II.1. The reads are displayed as pairs and sorted according to their insert size using IGV (Thorvaldsdottir, et al., 2013).

(B) Highlight of the WGS read pairs at the breakpoint junctions showing the typical drawing (read pairs pointing in opposite direction) for tandem duplications in direct orientation.

(C) Highlight of the WGS read pairs at the breakpoints including soft clipped bases. The left breakpoint in intron 30 overlaps the *AluJr* while the right breakpoint in intron 26 overlaps the *AluJb*. Analysis of the reads and the split reads that are misaligned allowed us to define the exact position of the duplication.



Figure S3. Homozygous duplication in *IFT140* validated by quantitative real-time PCR. Quantitative real-time PCR was performed on DNA from exon 30 in all individuals from family A and D as well as in one unrelated control. DNA quantity from exon 30 of *IFT140* was compared to 2 reference genes (*HBB* and *HMBS*) using the absolute quantification method.



Figure S4. Next generation sequencing data for family A and I displayed around *IFT140*, in particular from exon 27 to exon 30.

(A) Comparison between whole genome sequencing (WGS) from patient A-II.1 (tandem duplication carrier) and targeted exome sequencing using the full *IFT140* gene as a target region from patient I-II.1 (deletion of exon 27 to 29). The reads are displayed as pairs and sorted according to their insert size using IGV (Thorvaldsdottir, et al., 2013). The two events do not share the same breakpoints.

(B) Highlight of the read pairs from patient I-II.1 at the breakpoint junctions showing the typical drawing (read pairs pointing at each other) for a genomic deletion.

(C) Highlight of the read pairs from patient I-II.1 at the breakpoints including soft clipped bases. The right breakpoint overlaps a SINE element from the *Alu* family (Flam_C) but not the left breakpoint. Analysis of the split reads that are misaligned allows us to define the exact position of the deletion (c.3454-1005 4040+737delinsCCC).

	-	Chr 16		ما م	A-I.1	A-1.2	A-II.1*	A-II.2*	C-II.1*	B-II.2*	B-I.1	B-1.2	D-II.1*	Control 1	Control 2	Control 3	Control 4	Control !	Control 6
	rsiD	position	ret	alt		Dup	Dup	Dup	Dup	Dup	Dup								
	rs149678731	1560882	T	C	T/T 	T/T	T/T A/A	T/T 4/4	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T 4/4	T/T	T/T	T/C
	rs2667688	1561446	Â	G	A/A	A/G	A/A	A/A	A/A A/A	A/A A/A	A/G	A/A A/A	G/G	A/A	A/G	A/A	A/A A/A	A/G	A/A A/A
	rs572130902	1561488	G	с	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
	rs8048290	1562026	т	с	T/T	T/C	T/T	T/T	T/T	T/T	T/C	T/T	C/C	T/T	T/C	T/T	T/T	T/C	T/T
	rs12918013	1562195	A	G	A/A	A/G	A/A	A/A	A/A	A/A	A/G	A/A	G/G	A/A	A/A	A/A	A/A	A/G	A/A
	rs10083822 rs117143480	1562252	c	T	C/C	C/C	C/C	C/C	C/C C/C	C/C C/C	C/I	C/C C/C	1/1	C/C	с/т с/т	C/C	C/C C/C	C/T	C/C C/C
	rs8048980	1562460	т	c	т/т	T/C	т/т	т/т	T/T	T/T	T/C	T/T	c/c	т/т	T/C	т/т	т/т	T/C	T/T
	rs 1966171	1562804	G	А	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/A	A/A	G/G	A/A	G/G	G/A	G/A	G/G
	rs58369664	1562915	C	G	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/G	C/C	C/C	C/G	C/C	C/G	C/C	C/C
	rs112450135	1562987	G	C	G/G	G/C	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
	rs 10558385	1563667	ACTT	A	G/G	ACTIT/ACTT	ACTIT/ACTT	ACTITI/ACTI	G/G АСТТТ/АСТТ	G/G ACTIT/ACTI		G/G	A/A	G/G	ACTTT/A	ACTIT/ACTT	G/G ACTIT/ACTI		G/G
	rs113508951	1563672	С	СТ	C/C	C/C	C/C	C/C	C/C	c/c	C/C	C/CT	C/C	C/C	C/C	C/C	C/CT	C/C	C/C
	rs7191239	1563672	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
	rs67974359	1563705	T	C T	T/T	T/C	T/T	T/T	T/T	T/T	T/C	T/C	C/C	T/T	C/C	T/T	T/C	T/C	T/T
	rs 184085046	1563799	د ۵	6	L/L A/A	L/1 A/G	۵/۵	۲/C ۵/۵	C/C	C/C	C/1	C/C	1/1	L/L A/A	6/6	L/L A/A	C/C		C/C
	rs35773761	1563865	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/A	G/G
	rs551416387	1563876	G	А	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/G	G/G
	rs74430146	1563886	A	G	A/A	A/A	A/A	A/A	A/A	A/A	A/G	A/G	G/G	A/G	G/G	A/A	A/G	A/G	A/A
	rs62012850	1563896	C	A,G	C/C	C/G	C/C	C/C	C/C	C/C	C/G	C/A	G/G	C/C	C/C	C/C	C/A	G/G	C/C
	rs62012851	1563910	Ċ	т	C/C	C/T	C/C	C/C	1/1 C/C	1/1 C/C	Г/С С/Т	1/1 C/C	с/с т/т	C/C	C/C	C/C	1/1 C/C	і/с с/т	1/1 C/C
	rs2745111	1564823	G	А	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
	rs12597753	1565116	С	A	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/A	C/C	C/C	C/A	C/C	C/A	C/C	C/C
	rs11643891	1565540	C	T	C/C	C/T	C/C	C/C	C/C	C/C	C/T	C/T	T/T	C/C	T/T	C/C	C/T	C/T	C/C
	rs 79602039	1565893	A		A/A	Α/Α Δ/Δ	A/A	Α/Α Δ/Δ	A/A	A/A	A/A	A/1	A/A A/A	A/A	A/1 A/A	Α/Α Δ/Δ	A/1	A/A	A/A
	rs6600150	1566178	Â	c	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A
	rs752259914	1566178	А	ACC	A/A	A/A	A/A	A/A	A/A	A/A	A/ACC	A/ACC	ACC/ACC	A/A	A/A	A/A	A/ACC	A/ACC	A/A
	rs8051374	1566284	т	С	T/T	T/C	T/T	T/T	T/T	T/T	T/C	T/C	C/C	T/T	C/C	T/T	T/C	T/C	т/т
	rs112878182	1566444	A	G	A/A	A/G	A/A	A/A	A/A	A/A	A/A	A/A	A/A T/T	A/A	A/A	A/A	A/A	A/A	A/A
	rs2745185	1566597	т	c	т/т	т/с	т/т	т/т	T/T	T/T	T/C	T/C	c/c	т/т	C/C	т/т	T/C	T/C	T/T
	rs71385704	1566826	G	А	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/G	A/A	G/G	G/A	G/G	G/G	G/A	G/G
	rs150027092	1567367	AAG	A	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG	AAG/AAG
	rs147652844	1567368	AG	A	AG/AG	AG/AG	AG/AG	AG/AG	AG/AG	AG/AG	AG/AG	AG/AG	A/A	AG/AG	AG/AG	AG/AG	AG/AG	AG/A	AG/AG
	rs76268857	1567453	GAGA	G	AG/AG	AG/A	AG/AG GAGA/GAGA	AG/AG GAGA/GAG/	AG/AG	AG/AG				AG/AG	GAGA/G	GAGA/GAGA			
IFT140 Duplication	rs2072990	1567651	A	G	A/A	A/G	A/A	A/A	A/A	A/A	A/G	A/G	G/G	A/A	G/G	A/A	A/G	A/G	A/A
	rs2235637	1567694	G	A	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/A	A/A	G/G	A/A	G/G	G/A	G/A	G/G
	rs2076434	1568402	A	G	A/A	A/G	A/A	A/A	A/A	A/A	A/G	A/A	G/G	A/A	A/G	A/A	A/A	A/G	A/A
	rs /1385 /05 rs 117190102	1569003	A	۱ ۵	A/A G/G	A/1 G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G	A/T G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G	A/A G/G
	rs3784819	1569202	c	т	c/c	c/c	c/c	c/c	C/C	C/C	c/c	C/T	C/C	c/c	C/T	c/c	C/T	c/c	c/c
	rs3784820	1569252	G	А	G/G	G/A	G/G	G/G	G/G	G/G	G/A	G/G	A/A	G/G	G/A	G/G	G/G	G/A	G/G
	rs3784821	1569301	G	А	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
	rs3784822	1569308	C	T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
	rs199887622	1570059	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
	rs113734226	1570078	С	т	C/C	C/C	C/C	C/C	C/T	C/C	C/C	c/c	C/C	C/C	C/C	C/C	C/C	C/C	C/C
	rs932391	1570857	т	с	T/T	T/C	T/T	T/T	T/T	T/T	T/C	T/C	C/C	T/T	C/C	т/т	T/C	T/C	T/T
	rs932392	1571120	T	G	T/T	T/G	T/T	T/T	T/T	T/T	T/G	T/T	G/G	T/T	T/G	T/T	T/T	T/G	T/T
	rs111953232	1571811	G	GA	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
	rs9328919	1572282	С	т	C/C	C/T	C/C	C/C	C/C	C/C	C/T	c/c	T/T	C/C	C/T	C/C	C/C	C/C	C/C
	rs4787271	1572314	т	с	T/T	T/C	T/T	T/T	T/T	T/T	T/C	T/T	C/C	T/T	C/C	T/T	T/T	T/C	T/T
	rs9673971	1572393	G	A	G/G	G/A	G/G T/T	G/G	G/G	G/G	G/G	G/G	A/A	G/G	G/A			G/A	G/G
	rs76393829	1572594	Ċ	т	c/c	1/0	1/1 C/C	1/1 C/C	1/1	1/1	170.	7/0	C/C	7/7	cic	G/G	G/G	7/0	1/1
	rs554375875	1573219	1		0.0	U/L	C/C	C/C	C/C	C/C	C/C	T/C C/T	c/c	T/T C/C	C/C C/T	G/G T/T C/C	G/G T/C C/T	T/C	c/c
			CA	с	CA/CA	CA/CA	CA/CA	CA/CA	C/C CA/CA	C/C CA/C	C/C CA/CA	T/C C/T CA/C	c/c c/c c/c	T/T C/C CA/CA	C/C C/T CA/CA	G/G T/T C/C CA/CA	G/G T/C C/T CA/C	T/C C/C CA/C	C/C CA/CA
	rs11361072	1573219	CA CA	c c	CA/CA CA/CA	CA/CA CA/C	CA/CA CA/CA	CA/CA CA/CA	C/C CA/CA CA/CA	C/C CA/C CA/CA	C/C CA/CA CA/CA	T/C C/T CA/C CA/CA	C/C C/C C/C CA/CA	T/T C/C CA/CA CA/CA	C/C C/T CA/CA CA/C	G/G T/T C/C CA/CA CA/CA	G/G T/C C/T CA/C CA/CA	T/C C/C CA/C CA/CA	C/C CA/CA CA/CA
	rs11361072 rs34819117	1573219 1573485	CA CA G	C C A	CA/CA CA/CA G/G	C/C CA/CA CA/C G/G	C/C CA/CA CA/CA G/G	CA/CA CA/CA G/G	C/C CA/CA CA/CA G/G	C/C CA/C CA/CA G/G	C/C CA/CA CA/CA G/G	T/C C/T CA/C CA/CA G/A	C/C C/C C/C CA/CA G/G	T/T C/C CA/CA CA/CA G/G	C/C C/T CA/CA CA/C G/A	G/G T/T C/C CA/CA CA/CA G/G	G/G T/C C/T CA/C CA/CA G/A	T/C C/C CA/C CA/CA G/G	C/C CA/CA CA/CA G/G
	rs11361072 rs34819117 rs78028801 rs2745176	1573219 1573485 1573754 1573810	CA CA G C	C C A T G	CA/CA CA/CA G/G C/C G/G	C/C CA/CA CA/C G/G C/C	C/C CA/CA CA/CA G/G C/C	C/C CA/CA CA/CA G/G C/C G/G	C/C CA/CA CA/CA G/G C/C	C/C CA/C CA/CA G/G C/C	C/C CA/CA CA/CA G/G C/T	T/C C/T CA/C CA/CA G/A C/C	C/C C/C C/C CA/CA G/G C/T	T/T C/C CA/CA CA/CA G/G C/C G/G	C/C C/T CA/CA CA/C G/A C/T G/G	G/G T/T C/C CA/CA CA/CA G/G C/C G/G	G/G T/C C/T CA/C CA/CA G/A C/C G/G	T/C C/C CA/C CA/CA G/G C/T	C/C CA/CA CA/CA G/G C/C
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638	1573219 1573485 1573754 1573810 1573890	CA CA G C A G	C C T G A	CA/CA CA/CA G/G C/C G/G G/G	C/C CA/CA CA/C G/G C/C G/G G/G	CA/CA CA/CA G/G C/C G/G G/G	CA/CA CA/CA G/G C/C G/G G/G	C/C CA/CA CA/CA G/G C/C G/G G/G	C/C CA/C CA/CA G/G C/C G/G G/G	C/C CA/CA CA/CA G/G C/T G/G G/G	T/C C/T CA/C CA/CA G/A C/C G/G G/A	C/C C/C CA/CA G/G C/T G/G G/G	T/T C/C CA/CA CA/CA G/G C/C G/G G/G	C/C C/T CA/CA CA/C G/A C/T G/G G/A	G/G T/T C/C CA/CA CA/CA G/G C/C G/G G/G	G/G T/C C/T CA/C CA/CA G/A C/C G/G G/A	T/C C/C CA/CA G/G C/T G/G G/G	C/C CA/CA CA/CA G/G C/C G/G G/G
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437	1573219 1573485 1573754 1573810 1573890 1574030	CA CA G C A G	C C T G A C	CA/CA CA/CA G/G C/C G/G G/G G/G	C/C CA/CA G/G C/C G/G G/G G/G	CA/CA CA/CA G/G C/C G/G G/G G/G	CA/CA CA/CA G/G C/C G/G G/G G/G	C/C CA/CA G/G C/C G/G G/G G/G	C/C CA/CA G/G C/C G/G G/G G/C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G	T/C C/T CA/C CA/CA G/A C/C G/G G/A G/C	C/C C/C C/C CA/CA G/G C/T G/G G/G G/G	T/T C/C CA/CA G/G C/C G/G G/G G/G	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/A	G/G T/T C/C CA/CA CA/CA G/G C/C G/G G/G G/G	G/G T/C C/T CA/CA G/A C/C G/A G/A G/A	T/C C/C CA/CA G/G C/T G/G G/G G/G	C/C CA/CA CA/CA G/G C/C G/G G/G G/G
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038	1573219 1573485 1573754 1573810 1573890 1574030 1574170	CA G C A G G A	C A T G A C G	CA/CA CA/CA G/G C/C G/G G/G G/G A/A	C/C CA/CA G/G C/C G/G G/G G/G G/G A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A	C/C CA/CA G/G C/C G/G G/G G/G A/A	C/C CA/CA G/G C/C G/G G/G G/C A/A	C/C CA/CA G/G C/T G/G G/G G/G G/G	T/C C/T CA/CA CA/CA G/A C/C G/G G/A G/C A/A	C/C C/C C/C CA/CA G/G C/T G/G G/G G/G A/A	T/T C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G A/A	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G A/A	G/G T/C C/T CA/CA CA/CA G/A C/C G/G G/A G/G A/A	T/C C/C CA/CA G/G C/T G/G G/G G/G A/A	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235640	1573219 1573485 1573754 1573810 1573890 1574030 1574170 1574252 1574282	CA G C A G A A G	C A T G G G G	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/G G/G	C/C CA/CA G/G C/C G/G G/G G/G A/A A/G G/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G	C/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G A/A A/G	T/C C/T CA/CA CA/CA G/A C/C G/G G/A G/C A/A A/G G/C	C/C C/C C/C CA/CA G/G G/G G/G G/G G/G A/A G/G	T/T C/C CA/CA G/G C/C G/G G/G G/G A/A A/G G/G	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G G/A	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G	G/G T/C C/T CA/CA CA/CA G/A C/C G/G G/A G/G G/G G/G G/G	T/C C/C CA/CA G/G C/T G/G G/G G/G A/A A/A A/G	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235639 rs2235640 rs2235640 rs2745178	1573219 1573485 1573754 1573810 1573890 1574030 1574170 1574252 1574863 1575704	CA G C A G A G T	C A T G A C G A C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/G G/G C/C	C/C CA/CA G/G C/C G/G G/G G/G A/A A/G G/A C/C	C,/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C	C/C CA/CA G/G C/C G/G G/G A/A A/A G/G C/C	C/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/G C/C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G A/A A/G G/G C/C	T/C C/T CA/CA G/A C/C G/G G/G G/C A/A A/G G/G C/C	C/C C/C C/C CA/CA G/G C/T G/G G/G G/G G/G A/A G/G A/A C/C	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G G/A C/C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C	G/G T/C C/T CA/CA G/A C/C G/G G/A G/G G/G G/G G/G G/G C/C	T/C C/C CA/CA G/G G/G G/G G/G A/A A/G G/A C/C	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235640 rs2235640 rs2745178 rs112977910	1573219 1573485 1573754 1573810 1573890 1574030 1574170 1574252 1574863 1575704	CA G C A G A G T C	C A T G A C G A C T	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/G G/G C/C C/C	C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/G G/A C/C C/C	C,/C CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C	C/C CA/CA G/G C/C G/G G/G A/A A/A G/G C/C C/C	C/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/G C/C C/C	C/C CA/CA G/G C/T G/G G/G G/G A/A A/G G/G C/C C/C	T/C C/T CA/CA G/A C/C G/G G/A G/C A/A A/G G/G C/C C/C	C/C C/C C/C C/C G/G G/G G/G G/G G/G A/A G/G A/A C/C C/C	T/T C/C CA/CA G/G C/C G/G G/G G/G A/A A/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G A/A G/G G/A C/C C/C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C C/T	G/G T/C C/T CA/CA CA/CA G/A C/C G/A G/G G/A G/G G/G G/G C/C C/C	T/C C/C CA/CA G/G C/T G/G G/G G/G G/G A/A A/G G/A C/C C/C	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235640 rs2245178 rs112977910 rs2745181	1573219 1573485 1573754 1573754 1573810 1574030 1574030 1574170 1574252 1574863 1575704 1576858 1577056	CA G G G A G T C A	C A T G A C G A C T G	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G C/C C/C C/C	CA/CA CA/C G/G G/G G/G G/G A/A A/A G/A C/C C/C C/C C/C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C A/A	C/C CA/CA CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C	C/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/G C/C C/C C/C A/A	C/C CA/CA CA/CA G/G C/T G/G G/G G/G G/G A/A A/G G/G C/C C/C C/C A/G	T/C CT CA/C CA/CA G/A C/C G/A G/C A/A G/C A/A G/G C/C C/C C/C C/C	C/C C/C C/C C/C C/C G/G G/G G/G G/G A/A C/C C/C C/C G/G	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G A/A G/G G/A C/C C/C C/C C/C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A A/A G/G C/C C/T A/G	G/G T/C C/T CA/CA G/A C/C G/G G/A G/G G/A G/G G/G C/C C/C C/C C/C A/G	T/C C/C CA/CA G/G C/T G/G G/G G/G A/A A/G G/A C/C C/C C/C A/G	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235640 rs2745178 rs112977910 rs2745181 rs12598859 rs299859	1573219 1573485 1573754 1573810 1573890 1574030 1574170 1574252 1574863 1575704 1576858 1577056 1577184	CA G C A G G A A G T C A A	C A T G A C G G A C T G G	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G C/C C/C A/A A/A	CA/CA CA/CA G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C A/A A/C	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G C/C C/C A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A G/G C/C C/C A/A A/A	C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C A/G A/C	C/C CA/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/G C/C C/C C/C A/A A/A	C/C CA/CA CA/CA G/G G/G G/G G/G A/A A/G G/G C/C C/C C/C C/C A/G A/A	T/C C/T CA/CA G/A C/C G/A G/C A/A A/G G/C A/A C/C C/C C/C C/C A/G A/G	C/C C/C C/C C/C C/T G/G G/G G/G A/A G/G A/A G/G A/A C/C C/C C/C C/C C/C C/C	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G A/A G/G G/A C/C C/C C/C C/C G/G A/G	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A A/A A/A A/A C/C	G/G T/C C/T CA/CA G/A C/C G/G G/A G/G G/A G/G G/G A/A G/G C/C C/C C/C C/C A/G A/G	T/C C/C CA/CA G/G C/T G/G G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C A/G A/A	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C A/A A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235639 rs2235640 rs2745178 rs112977910 rs2745181 rs1259859 rs939655 rs939655	1573219 1573485 1573754 1573810 1573890 1574030 1574170 1574252 1574863 1575704 1576858 1577056 1577184 1577963 1577963	CA GC GG GA GG A G CA A A C	C A T G A C G G G G T G G T	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C A/A A/A A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A	C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A C/C C/C C/C A/A A/A A/A C/C	C/C CA/CA G/G G/G G/G G/G G/C A/A A/A A/A A/A A/A A/A A/A A/A	C/C CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A A/G C/C C/C C/C C/C A/A A/A A/A	T/C C/T CA/CA G/A C/C G/A G/A G/C A/A A/G G/G C/C C/C C/C A/G A/G C/C	C/C C/C C/C CA/CA G/G G/G G/G G/G A/A G/G A/A G/G C/C C/C C/C C/C C/C	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C A/A A/A A/A A/A A/A C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G G/A G/G G/A C/C C/C C/C G/G A/G G/G C/C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G A/A A/A A/A A/A A/A	G/G T/C C/T CA/CA G/A C/C G/A G/G G/A G/G G/A G/G G/G C/C C/C C/C C/C	T/C C/C CA/CA G/G C/T G/G G/G G/G G/G G/G A/A A/G C/C C/C C/C C/C A/G A/A A/G C/T	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C A/A A/A A/A A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs2235638 rs2076437 rs141171038 rs2235640 rs2745178 rs122977910 rs2745181 rs12599859 rs939855 rs192452222 rs140400369	1573219 1573485 1573754 1573810 1574030 1574030 1574030 1574252 1574863 1575704 1576858 1577056 1577184 1577963 15778295 1578373	CA CA G C A G G A A G T C A A C C	C A T G A C G G G G T G G T G	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/G G/G C/C C/C C/C A/A A/A A/A A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A	C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C	C/C CA/CA G/G C/C G/G G/C A/A A/A A/A A/A A/A A/A A/A A/A A/A A	C/C CA/CA CA/CA G/G C/T G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C	T/C C/T CA/CA G/A C/C G/G G/A G/C A/A A/G G/G C/C C/C C/C A/G A/G A/G C/C C/C	C/C C/C C/C G/G G/G G/G G/G G/G G/G G/G	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/G G/A C/C C/C C/C C/C C/C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G G/G C/C C/T A/G A/A A/A A/A A/A C/C C/T C/C	G/G T/C C/T CA/CA G/A C/C G/A G/G G/A G/G G/G G/G G/G C/C C/C C/C A/G A/G A/G A/G C/C C/C	T/C C/C CA/CA G/G G/G G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A
	s11361072 s34819117 s78028801 s2745176 s2235638 s2076437 s141171038 s2235639 s2235639 s2235639 s122375178 s112977910 s2745178 s112599859 s19239655 s192452222 s140400369	1573219 1573485 1573754 157380 157380 1574030 1574030 1574252 1574863 1575704 1576858 1577056 1577184 1577963 1578295 1578373 1578434	CA GC GG GA GG A A G C G G G	C C A T G A C G G A C T G G G G G G C A	CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A A/	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A A/	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/C G/G G/C G/C A/A A/A A/A A/A A/A A/A A/A A/A A/A C/C C/C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G G/G C/C C/C C/C C/C C/C C/C	T/C C/T CA/CA G/A C/C G/A G/C G/A G/C C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C C/C C/C C/C C/T G/G G/G G/G A/A C/C C/C G/G G/G A/A G/G C/C C/C C/C GCA/GCA	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/CA CA/CA CA/C G/A C/T G/G G/A G/G A/A G/G G/A C/C C/C C/C G/G G/G C/C C/C C/C G/CA/CA	G/G T/T C/C CA/CA G/G C/C C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/G C/C C/C C/C C/C	G/G T/C C/T CA/C G/A G/A C/C G/A G/G G/G G/G G/G C/C C/C C/C C/C A/G A/G A/G C/C C/C C/C C C/C	T/C C/C CA/CA G/G C/T G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A A/A A/A C/C C/C C/C C/C C/C C/C
	s11361072 s34819117 s78028801 s2745176 s2235638 s2235638 s2235639 s2235639 s2235639 s2235640 s2745178 s112977910 s2745181 s1259859 s192452222 s140400369 s149176109 s14927610	1573219 1573485 1573754 1573810 1573890 15747030 15747252 1574863 1577056 1577184 1576858 1577056 1577184 1577633 1578295 1578373 1578374 1578514	CA CA G C A G G A A G T C A A A C C G A G	C C A T G A C G G A C T G G C C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A A/	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A A/	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/C G/G G/G G/C A/A A/A G/G C/C C/C C/C C/C A/A A/A A/A A/A A/A C/C C/C	C/C CA/CA G/G C/T G/G G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	T/C C/T CA/CA G/A C/C G/A G/A G/C C/C C/C C/C C/C A/G A/G C/C C/C C/C G/GCA A/C	C/C C/C C/C C/C C/C C/C G/G G/G G/G G/G	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/C G/A G/G G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G G/G C/C C/T A/G A/A A/A G/G C/C C/T A/G C/C C/C C/C C/C	G/G T/C C/T CA/CA G/A C/C G/A G/A G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A A/A C/C C/C C/C C/C C/C C/C C/C
	s11361072 s34819117 s78028801 s2745176 s2235638 s2076437 s141171038 s2235639 s2235639 s2235640 s2745178 s112599859 s5939655 s192452222 s140400369 s149176109 s1983601 s1983601 s1983601 s1983601	1573219 1573485 1573754 1573754 1573810 1574800 1574030 1574030 1574252 1574863 1575704 1576858 1577058 1577058 15778434 1578373 1578434 1578514	CA GCAGCAGGAAGTCAAACCGAG▲	C C A T G A C G G A C T G G T G GC A C	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G C/C C/C A/A A/A C/C C/C C/C A/A A/A C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C G/GCA A/A G/G G/GCA	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C A/A A/A C/C C/C	CA/CA CA/CA G/G C/C G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C A/A A/A G/G G/G A/A	C/C CA/CA G/G G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C G/GCA A/A G/G A/A	C/C CA/CA CA/CA CA/CA C/C G/G G/C A/A A/A C/C C/C C/C C/C C/C C/C C/C C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C A/A A/G C/T C/G G/GCA A/A G/G G/G	T/C CA/CA CA/CA G/A C/C G/A G/G G/G G/C A/A A/G A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C C/C C/C CA/CA G/G G/G G/G A/A G/G A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G C/C A/A A/A C/C C/C C/C A/A A/A C/C C/C	C/C C/T CA/CA CA/C G/A C/C G/A G/G G/A G/G G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/T A/G A/A C/C C/C C/C C/C C/C C/C G/GCA A/A G/G	G/G T/C C/T CA/CA G/A C/C G/A G/A G/G G/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/C G/G G/G G/G G/G G/G G/G G/G G/A C/C C/C C/C C/C C/C C/C C/T C/G G/GG A/A A/A A/A A/A A/A A/A	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A
	rs11361072 rs34819117 rs78028801 rs2745176 rs225638 rs225638 rs2256437 rs141171038 rs2235639 rs2235640 rs2235640 rs2245178 rs112977910 rs2745178 rs112977910 rs2745178 rs112977910 rs2745178 rs1298850 rs192452222 rs19245222 rs192452 rs19452 rs1957 rs1957 rs1957 rs195757 rs	1573219 15732485 1573754 1573754 1573800 1574030 1574030 1574252 1574863 1577055 1577705 1577755 15775	CA CA G C A G G A A G T C A A A C C G A G A G	C C A T G A C G G A C T G G T G GA G A G A G A G A G A G A G	CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C A/A A/A C/C C/C C/C A/A A/A C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C A/A A/A C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A A/A A/A A/A	C/C CA/CA G/G C/T G/G G/G G/G G/G C/C C/C C/C C/C C/C C/C	T/C C/T CA/CA G/A C/C G/G G/G G/G G/C A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C A/G A/G A/G A/G C/C C/C C/C C/C A/CA	C/C C/C C/C C/C C/C C/C C/C G/G G/G G/G	T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA G/A C/T G/G G/A G/G G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C C/C C/C C/C	G/G T/C C/T CA/CA G/A C/C G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/C G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C A/G A/A A/G C/T C/G G/GCA A/A A/G G/G A/A	C/C CA/CA G/G G/G G/G G/G G/G A/A A/A A/A A/A A/
	s11361072 s34819117 s78028801 s2745176 s2235638 s2076437 s124171038 s2235640 s2235640 s2235640 s2235640 s2245178 s112977910 s2745178 s1259939655 s192452222 s140400369 s1938000 s1938000 s1938000 s1938000 s1938001 s19384825 s138773419 s138773419	1573219 15732485 1573754 1573810 1573830 1574030 1574030 1574030 1574030 1574030 1574052 1574053 1576858 1577846 1577056 1577184 1577056 1577184 1577855 1578373 1578834 1578834 1578834	CA CA G C A G G A A G T C A A A C C G A G A G T	C C A T G A C G G A C T G G G T G G A C G G A C T G G A C T G G A C A G A C	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	CA/CA CA/C G/G G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C G/GCA A/A A/G G/G A/A A/G G/G A/A	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G A/A A/A A/A A/A A/A	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G A/A A/A A/A A/A A/A	C/C CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/C A/A A/A A/A C/C C/C C/C C/C C/C C/C G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA CA/CA G/G C/T G/G G/G G/G G/G G/G C/C C/C C/C C/C A/A A/G C/C C/C A/A A/G G/GCA A/A G/G G/GCA A/A G/G G/GCA	T/C C/T CA/CA G/A C/C G/A G/A G/C A/A G/C C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C C/C C/C C/C C/T G/G G/G A/A G/G C/T C/C G/G A/A G/G C/C C/C C/C G/G A/A G/G C/C C/C C/C C/C C/T	T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	C/C C/T CA/CA CA/C G/A C/T G/G G/A G/A G/A G/A G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/T A/A C/C C/C C/T A/A C/C C/C G/GCA A/A A/A G/G C/C C/C T/T	G/G T/C C/T CA/CA G/A C/C G/A G/A G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA G/G G/G G/G G/G G/G A/A G/A C/C C/C C/C C/C A/A A/G G/GCA A/A G/G G/GCA A/A G/G G/GCA	C/C CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C A/A A/A A/A C/C C/C G/G A/A A/A G/G C/C
	s11861072 s34819117 s78028801 s2745176 s2235638 r22056437 r241171038 r2235640 r2745178 s112977910 s2745181 r512598859 r5192452222 r5140400365 r51983600 r51983601 r51984601 r51983601 r51984601 r51983601 r51984601 r51984601 r51984601 r51984601 r51984601 r51984601 r519846000 r519846000 r519846000000000000000000000000000000000000	1573219 15732485 1573754 1573754 1573810 1574030 1574030 1574030 1574050 1574050 1574050 1574050 1574050 1574050 1577053 1577053 1577053 1577053 1577053 1577053 1578053 1578054 15780554 1578054 1578054 1578054 1578054 1578054 1578054 1578	CA C G C A G G A A G T C A A A C C G A G A G T C	C C A T G A C G G A C T G G G T G A C A G A C T	CA/CA CA/CA G/G C/C G/G G/G G/G G/G G/G G/G C/C C/C	CA/CA CA/CA CA/C G/G C/C G/G G/G G/G G/G G/A A/A C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C G/G A/A A/A C/C C/C G/G A/A C/C C/C C/C C/C C/C	CA/CA CA/CA G/G C/C G/G G/G A/A A/A A/A C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A C/C C/C C/C C/C C/C G/GCA A/A G/G A/A G/G T/T T/C/C	C/C CA/CA G/G G/C G/G G/C A/A A/A G/G C/C C/C C/C C/C C/C C/C G/G A/A A/A C/C C/C G/G A/A C/C	C/C CA/CA G/G C/T G/G G/G G/G G/G C/C C/C C/C C/C A/A A/G C/T C/G G/GCA A/A G/G T/T T/C/C	T/C C/T CA/CA G/A G/A G/A G/C G/A G/A G/C C/C C/C C/C C/C C/C C/C C/C C/C G/GCA A/G A/G A/G A/C G/A C/C C/C C/C C/C C/C C/C C/C	C/C C/C C/C CA/CA G/G G/G C/T G/G G/G A/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/T C/C CA/CA G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	C/C C/C CA/CA G/A C/T G/A C/T G/G G/A G/A G/A G/A G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/T A/A A/A A/A A/A A/A C/C C/C C/C C/C C/C	G/G T/C C/T CA/CA G/A G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA G/G C/T G/G G/G G/G A/A G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C G/GCA A/A G/G A/A G/G T/T T/C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A A/A A/A A/A A/A A/A A/A
	s11861072 s34819117 s78028801 s2745176 s2235638 s2745176 s2235640 s2745178 s2745178 s129793005 s129779100 s129793005 s1297745181 s12979800 s149176109 s149476109 s149476109 s1494822222 s140400369 s149476109 s1494822222 s140400369 s149476109 s1494822222 s140400369 s149476109 s149482222 s140402491 s3748428 s138774419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3748428 s138784419 s2064291 s3784828 s14881225 s748828 s1488828 s1488828 s1488828 s14888 s148888 s148888 s148888 s1488888 s1488888 s1488888 s148888 s148888 s148888 s148888 s1488888 s1488888 s1488888 s1488888 s1488888 s1488888 s1488888 s1488888 s148888 s1488888 s1488888 s1488888 s1488888 s1488888 s1488888 s1488888 s14888888 s14888888 s14888888 s14888888 s14888888 s14888888 s14888888 s14888888 s148888888 s148888888 s1488888888888 s1488888888888 s148888888888888888 s14888888888888888888888888888888888888	1573219 1573254 1573754 1573754 1573810 1574270 1574120 1574120 1574120 1574252 1574563 1577056 1577184 1578553 1578295 1578295 1578295 1578295 1578295 1578295 1578295 1578295 1578295 1578294 1578295	CA C G C A G G A A G T C A A A C C G A G A G T C T -	C C A T G A C G G A C T G G G T G GC A G A C T C	CA/CA CA/CA G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/A A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A A/A A/A C/C C/C C/C G/G A/A A/A C/C C/C C/C G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A A/A A/A A/	C/C CA/CA G/G G/G G/G G/G G/G A/A A/A A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C CA/CA G/G G/G G/G G/G G/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/T G/G G/G G/G G/G C/T C/C C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/T CA/CA G/A G/A G/A G/A G/A A/G G/A A/G C/C C/C C/C C/C G/GCA A/G A/G C/C C/C G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C C/C C/C C/C C/C C/C G/G G/G G/G G/G	T/T C/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C G/G C/C C/C	C/C CT CA/CA CA/CA CA/CA C/T G/A C/C G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G C/C CT A/A A/G C/C C/T A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/C CA/CA G/A G/A G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA G/G G/G G/G G/G G/G A/A A/G G/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C A/A A/G G/G G/G A/A G/G A/A	C/C CA/CA CA/CA G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C
	s11861072 s34819117 s78028801 s2745176 s2235638 s2745176 s2235639 s2235640 s2745181 s12598899 s2235640 s2745181 s125998899 s129393655 s192462222 s140400369 s1384074109 s138407410 s138477419 s138773419 s138775 s138755 s138755 s138755 s138755 s138755 s1387555 s1387555 s13875555 s1387555555555555555555555555555555555555	1573219 1573254 1573754 1573754 1573810 1574750 1574252 1574030 1574252 1574030 1574252 1574030 1574252 1574043 1577063 1577063 1577063 1577063 1577054 1578254 1579254 15795554 157955555555555555555555555555555555555	CA CG CA GCA GGAAGTCAAACCGAGAGTCTC▲	C C A T G A C G G A C T G G G T G G C A G A C T C T G	CA/CA CA/CA G/G G/G G/G G/G G/G C/C C/C A/A A/A C/C C/C C/C C/C A/A A/A	CA/CA CA/CA CA/C G/G G/G G/G G/G A/A C/C C/C C/C C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C A/A A/A C/C C/C G/G A/A A/A C/C C/C G/G A/A	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C A/A A/A C/C C/C C/C A/A A/A C/C C/C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G G/C G/C G/C A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/T G/G G/G G/G G/G G/G C/T C/C C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/T CA/CA G/A C/C CA/CA G/A G/G G/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	С/С С/С С/С С/С С/С С/С С/С С/С С/С С/С	T/T C/C CA/CA G/G C/C G/G G/G G/G G/G G/G C/C A/A A/A C/C C/C C/C G/G A/A A/A C/C C/C C/C G/G A/A A/A C/C C/C C/C C/C A/A A/A C/C C C/C A/CA A/A A/	C/C C/C CA/CA CA/CA CA/C G/A C/T G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/T A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	G/G T/C C/T CA/CA G/A G/G G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A C/C C/C C/C C/C C/C C/C C/C C/C C/C	C(C CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C
	s11361072 s74819117 s78028801 s2745176 s2235638 r22056437 r141171038 r2235640 r2745178 s112977910 s2745178 r192452222 r132773419 r512460025 r149176109 r51983600 r51983601 r51984602 r5198	1573219 15732485 1573754 1573754 1573810 1574030 1574030 1574030 1574023 1574853 1574554 1577653 1577653 1577653 1577653 15777184 1578254 1578254 1578254 1578999 1579151 1579151 1579244 1579454 1579448 15794445 1579445 157945 157945 157945 157945 157945 157945 157945 157945 157945 157945 157945 157945 1579745 1579745	CA G G C A G G A A G T C A A A C C G A G A G T C T C A G	C C A T G A C T G G G A C T G G A C T G G A C T G G A C T G G A C T G A C T C T G A	CA/CA CA/CA G/G G/G G/G G/G G/G C/C C/C A/A A/A C/C C/C C/C C/C C/C A/A A/A	CA/CA CA/CA CA/C G/G G/G G/G G/G G/G G/G C/C C/C C/C C	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C G/G A/A A/A C/C C/C G/G A/A A/A C/C C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G C/C G/G G/G G/G A/A A/A G/G C/C C/C C/C A/A A/A C/C C/C C/C G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C	C/C CA/CA G/G C/T G/G G/G G/G A/A A/G C/C C/C C/C C/C C/C C/C C/C C/C C/T C/T	T/C C/T CA/CA G/A C/C G/A G/A G/C G/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C C/C C/C C/C C/C C/C C/C C/C C/C C/C	T/T C/C CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	C/C C7/C CA/CA CA/CA C/T G/A G/A G/A G/A G/A G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G G/G C/C C/T A/A A/A C/C C/C C/T G/GA/A A/A G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	G/G T/C CA/CA G/A G/A G/G G/A G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA CA/CA CA/CA C/T G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	C/C CA/CA CA/CA G/G G/G G/G G/G A/A A/A A/A A/A A/A C/C C/C C/C C/C G/G A/A A/A A/A G/G G/G A/A C/C C/C C/C C/C C/C C/C C/C C/C A/A
	s11861072 s34819117 s78028801 s2745176 s2235638 s2235640 s2235640 s2235640 s2745178 s1129793100 s12973610 s12973610 s12973610 s12983600 s1983600 s1983600 s1983600 s19836221 s19834255 s1983455	1573219 1573254 1573754 1573754 1573810 1574270 1574170 1574170 1574252 157463 1574252 1574563 1577056 1577154 1577056 1577154 1578295 1578295 1578295 1578295 1578295 1578291	CA G C G C A G G A A G T C A A A C C G A A G T C A A G G T C A G G T C A G G C A G C A G C C A G C A G C A G C A G C C A G C C A G C C A G C C A G C C C A G C C C C C C C C C C C C C C C C C C C	C C A T G A C G G G G G G G C A C T C T G A C T C T G A C T C T G A T	CA/CA CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	C/C CA/CA CA/CA G/G C/C G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G C/C G/G G/G C/C A/A G/G C/C C/C A/A A/A C/C C/C C/C A/A A/A C/C G/G G/G T/T C/C C/C A/A A/A G/G	C/C CA/CA CA/CA G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C	T/C C/T C/C C/C CA/CA G/A G/A G/C G/C C/C C/C C/C C/C C/C C/C C/C C/C	С/С С/С С/С С/С С/С С/С С/С С/С С/С С/С	Т/Т С/СС СА/СА СА/СА G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	C/C CA/CA CA/CA CA/CA C/C G/A G/A G/A G/A G/A G/A G/A G/A C/C C/C C/C C/C C/C C/C C/C C/C GCA/GCA A/A G/A G/A G/A G/A G/A G/A G/A G/A G/	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C G/CC A/A A/A A/A G/G G/C A/A G/G A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	G/G T/C C/T CA/CA G/A C/C G/A G/A G/G G/A G/G G/G C/C C/C C/C C/C C/C C/C G/GCA A/G A/G A/G A/G A/G C/C C/C C/C C/C C/C C/C C/C C/C C/C A/CA G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C	T/C C/C CA/CA CA/CA CA/CA CA/CA C/C C/C C	C/C CA/CA G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C
	s11861072 s34819117 s78028801 s2745176 s2235638 s2745176 s2235639 s2235640 s2745178 s112977910 s2235640 s2745181 s129939655 s192452222 s140400269 s1938001 s3784825 s138773419 s1294745183 s138773419 s138774518 s13877455555555555555555555555555555555555	1573219 1573254 1573754 1573754 1573810 1574252 1574170 1574252 1574270 1574252 1574252 1574252 1574252 1574253 1577554 1577554 1577554 1578514 1578514 1578514 1578514 1578914 15789151 1579151 1579255 1578925 1578925 1578925 1578925 1578925 1578925 1578925 1578925 1578925 1578925 1578925 1580752 1580752	CA G C A G G A A G T C A A A C C G A G T C T C A G G G A	C C A T G A C G G G G T G G A C T C T G A T G G C A C T C T G A T G G C A C T C T G A T G	CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A A/A A/A C/C C/C C/C C/C C/C C/C	CA/CA CA/CA CA/C G/G G/G G/G G/G G/G G/G A/A C/C C/C C/C C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C C/C C/C C/C	CA/CA CA/CA G/G G/G G/G G/G G/G G/G A/A A/A A/A C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA CA/CA G/G G/G G/G G/G G/G A/A A/A C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/G G/G G	T/C C/T CA/CA G/A G/A G/C G/G G/A G/C C/C C/C C/C C/C C/C G/GC A/G A/G C/C G/GC A/G G/A T/C G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	С/С С/С С/С С/С С/С С/С С/С С/С С/С С/С	Т/Т С/С СА/СА СА/СА (G)G С/С G)G G)G G)G G)G G)G G)G G)G C/C C/C C/C C/C C/C C/C C/C C/C C/C C/	Cr.C Cr.C Cr.C CA/CA CA/CA G/G G/A G/G G/A G/G G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G G/G G/G G/G A/A A/A G/G C/C C/T A/G C/C C/T C/T C/T C/T T/T T/T C/T T/T C/T T/T C/T A/A A/A	GiG TIC CT/C CA/CA GIA GIA GIG GIG GIG GIG GIG GIG C/C C/C C/C C/C C/C C/C C/C C/C C/C C/	T/C C/C CA/CA CA/CA CA/CA G/G G/G G/G G/G G/G G/G C/C C/C A/A C/C C/C A/A A/A G/A C/T C/G G/GC C/C C/C A/A A/A G/C C/C C/C C/C C/C C/C C/C C/C C/C C/C	C/C CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C
	s11861072 s34819117 s78028801 s2745176 s2235638 s2235640 s2235640 s2235640 s2235640 s2235640 s2745178 s112977910 s2745181 s12598850 s1938600 s1938601 s1383601 s1383601 s1383601 s1383601 s1383601 s1383601 s1383601 s1383601 s138374428 s138374428 s138374429 s13837454555555555555555555555555555555555	1573219 15732485 1573754 1573754 1573830 1574030 1574030 1574030 1574030 1574030 1574030 1574030 1574030 1574056 1577184 1577805 1577184 1577855 1577184 15778514 157825 1578373 1578514 157825 1578373 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 1578483 1578514 157852 1578514 157852 1578514 157852 158752 1587752 1587770 1582870 1582870	CA G G C A G G A A G T C A A A C C G A G G A G G G A G G G A C C C C	C C A T G A C G G A C T G G G T G GCA C A C T G A T G T G T G T G T G T G T G T G T	CA/CA CA/CA GA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	CA/CA CA/CA CA/C G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	С.С. А/С.А С.А/С.А С.А/С.А С.А/С.А С.А/С. С.G.G G.G.G G.G.G G.G.G G.G.G C.C C.C C.C	С.С. А/СА С.А/СА С.А/СА С.А/СА С.А/СА С.С. С.С.	C/C	C/C CA/C CA/CA G/G G/C G/C G/C G/C G/C G/C C/C C/C C/C	С/С СА/СА СА/СА СА/СА С/Г С/Г С/Г С/Г С/Г С/С С/С С/С С/С С/С	T/C C/T C/A/CA G/A C/C G/A G/A G/C G/G C/C C/C C/C C/C C/C C/C C/C C/C		Т/Т С/С СА/СА/СА СА/СА/СА СА/СА/СА/СА/СА СА/СА/СА/СА СА/СА/СА/СА/СА/СА/СА/СА/СА/СА/СА/СА/СА/С	CrC CrC CrC CA/CA CA/CA CA/CA CrC G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	G/G T/T C/C CA/CA G/G G/G G/G G/G G/G A/A A/A G/G C/C C/T A/A A/A C/C C/C G/GCA A/A G/G A/A G/G A/A G/G A/A G/G A/A G/G A/A G/G A/A C/C C/C C/C C/C C/C C/C C/C C/C C/C	GiG GIG TIC CA/C CA/CA GIA GIG GIG GIG GIG GIG GIG GIG GIG C/C C/C C/C C/C C/C C/C C/C C/C C/C C/	ТСС С/С С/С С/С С/С С/С С/С С/С	C/C C/C CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C A/A A/A A/A G/G G/G G/G G/G A/A A/A A/A A/A G/G G/G G/G A/A A/A A/A A/A
	s11861072 s34819117 s78028801 s2745176 s2235638 s2235640 s2235640 s2235640 s2745178 s112979100 s7745181 s1129798100 s7745181 s1129798100 s745178 s1129798100 s149176109 s149176100000000000000000000000000000000000	1573219 1573254 1573254 1573754 1573810 1574270 1574270 1574270 1574252 1574252 1574252 1574252 1574252 1574252 1577154 1577056 1577154 1577054 1578255 1578254 15782577 15882577 15882577 15885777 15885777 15885777 15885777 158857777777777	CA G C A G G A A G T C A A A C C G A G A G T C T C A G G A C C A	C C A T G A C G G A C T G G G T G GCA G A C T C T G A T G T T .	CA/CA CA/CA GA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	СС СА/СА СА/СС СА/СС СА/СС С/С С/С С/С С	C/C A CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	C) C A/CA CA/CA CA/CA G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C	CC/C CA/CA G/G G/G G/G G/G G/G G/G G/G A/A A/A A/	С/С СА/С СА/С С/С С/С С/С С/С С/С С/С С/	С/С СА/С СА/СА СА/СА С/Т С/Т С/Т С/Т С/С С/С С/С С/С	T/C C/T C/C C/C CA/CA G/A C/C G/A G/A G/C C/C C/C C/C C/C C/C C/C C/C C/C C/C	С/С С/С С/С С/С С/С С/С С/С С/С С/С С/С	Т/Т СА/СА СА/СА СА/СА СА/СА СА/СА СА/СА СА/СА СА/СА СА/С СА/С СА/С СА/С А/А А/А	Cr.C Cr.C Cr.C CA/CA CA/CA CA/CA Cr.C C/C C/C C/C C/C C/C C/C C/C C/C C/C C	G/G T/T C/C CA/CA G/G C/C G/G G/G G/G G/G A/A A/A G/G C/C C/C C/C C/C C/C G/GCA A/A A/A G/G G/G A/A G/G A/A G/G G/G A/A G/G C/C C/C C/C C/C C/C C/C C/C C/C C/C	Gig TiC Cr CA/CA G/A G/A G/G G/G G/G G/G G/G G/G G/G G/	T(C C)C C/C C/C C/C C/C G/G G/G G/G G/G G/G G/	С/С С СА/СА G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C

Page 46	of 52
---------	-------

	rs10576	512 1	584446	G	С	G/C	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	L
	rs20764	38 1	584618	т	С	T/C	T/C	T/T	T/T	T/C	T/T	T/C	T/C	C/C	T/T	C/C	T/C	C/C	T/C	T/T	
	rs12446	975 1	584628	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs 14383	3421 1	584866	т	ĉ	T/C	T/C	T/T	T/T	G/G T/C	G/A T/T	G/A T/C	G/G T/C	G/G	G/G T/T	C/C	T/C	G/G	G/G T/C	G/A T/T	
	rs11690	3565 1	584867	G	Ā	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/G	G/G	
	rs22356	543 1	585115	G	А	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G	
	rs71966	49 1	585456	A	G	A/G	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	
	rs1414t	105 1	585650	C	т	A/G C/T	A/A C/C	A/G C/C	A/G C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A C/C	
	rs22946	24 1	586255	G	Å	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs 19026	55545 1	586275	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/C	C/C	C/C	C/C	
	rs22946	25 1	586596	С	Т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C	
	rs 22946	26 1	586/1/	G	G	C/C	6/6	6/6	C/C G/G	C/G	C/C	C/G	C/C	G/G	6/6	C/G G/A	6/6	C/G G/A	C/G	C/C G/G	
	rs12923	143 1	587067	т	c	т/т	т/т	т/т	т/т	T/T	T/T	т/т	T/T	T/T	т/т	т/т	т/т	т/т	T/C	T/T	
	rs20764	39 1	587277	С	т	C/T	C/C	C/C	C/C	C/T	C/C	C/T	C/T	T/T	C/C	T/T	C/T	T/T	C/T	C/C	
	rs20764	40 1	587322	G	A	G/A	G/G	G/G	G/G	G/A	G/G	G/A	G/A	A/A	G/G	A/A	G/A	A/A	G/A	G/G	
	rs 20665	15747 1	587511	G	т	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/A C/C	G/G C/C	G/A C/C	G/G C/C	A/A C/C	G/G C/C	G/A C/T	G/A	G/A C/C	G/A C/C	G/G C/C	
	rs78140	0717 1	587938	c	т	C/C	c/c	c/c	c/c	c/c	c/c	c/c	C/T	c/c c/c	c/c	С/Т	c/c	C/T	c/c	c/c	
	rs80512	95 1	588087	А	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A	
	rs 28668	12520 1	588800	G	A	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G	
	rs 18980	17580 1	588849 589671	т	Ċ	C/C T/T	C/C T/T	с/с т/т	с/с т/т	T/T	U/C T/T	T/T	с/с т/т	T/T	T/C	T/T	T/T	T/T	T/T	C/C T/T	
	rs80113	230 1	590168	G	Ā	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs11694	1519 1	590480	G	А	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	
	rs20764	41 1	590576	С	G	C/G	C/G	C/G	C/G	C/G	C/G	G/G	C/G	G/G	C/C	G/G	C/G	G/G	G/G	C/G	
_	rs66001 rs20764	42 1	590672 591112	G	A T	G/A G/T	G/G	G/G	G/G	G/G G/T	G/G G/G	G/G G/T	G/A G/T	G/G T/T	G/G	G/A T/T	G/G	G/A T/T	G/G G/T	G/G G/G	
	rs11718	9215 1	591284	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	L
	rs 20501	17 1	591592	G	А	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G	
	rs20764	43 1	591962	G	A	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G	
	rs20764	44 1	592087		т	A/T	1/1 A/A	1/1 A/A	1/1 A/A	1/C	1/T	1/C	1/T	C/C T/T	1/1 A/A	T/T	1/C A/T	T/C	1/C	1/T	
	rs11649	407 1	592367	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs20764	46 1	592524	С	т	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	T/T	C/C	C/T	C/T	C/T	C/T	C/C	
	rs 37848	30 1	592857	G	c	G/C	G/G	G/G	G/G	G/G	G/G	G/G	G/C	G/G	G/G	G/C	G/G	G/C	G/G	G/G	
_	rs 20501	132 1	593108	G	A C	G/G G/C	G/G	G/G	G/G	G/A	G/G	G/A	6/6	A/A G/G	G/G	G/A G/C	G/A G/G	G/A G/C	G/A	G/G	
	rs21432	85 1	593171	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A	1
	rs80499	972 1	593606	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C	
	rs 20642	89 1	593645	C	T	C/T	C/C	c/c	c/c	C/T	C/C	C/T	C/T	T/T	C/T	T/T	C/T	T/T	C/T	C/C	L
	rs45584 rs80536	18 1 505 1	593843 594158	G	A	G/G G/A	G/G	G/G	G/G	G/A G/G	G/G G/G	G/A G/A	G/G G/A	A/A A/A	G/G	G/A A/A	G/A G/A	G/A A/A	G/A G/A	G/G G/G	
	rs80536	08 1	594161	G	т	G/G	G/G	G/G	G/G	G/T	G/G	G/G	G/G	T/T	G/G	G/T	G/T	G/T	G/T	G/G	
	rs56978	84189 1	594162	GCACCTGO	G	CTGC/GCAC	CTGC/GCAC	CTGC/GCAC	CTGC/GCAC	CTGC/GCAC	CTGC/GCA	CCTGC/GCAC	SCACCTGC/	CTGC/GCAC	CTGC/GCAC	CTGC/GCAC	CCTGC/GCA	GCACCTGC/	CTGC/GCAC	CTGC/GCA	
	rs15067	2825 1	594218	T	A	T/A	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	
	rs 35055	895 1	594248 594314	A	G	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/G A/G	A/G	A/A A/A	A/A A/A	A/A A/A	
	rs14018	9706 1	594376	т	c	T/C	т/т	т/т	т/т	т/т	т/т	т/т	т/т	т/т	т/т	т/т	т/т	т/т	T/T	т/т	
	rs 19206	8259 1	594672	т	с	T/C	T/T	T/C	T/C	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	
	rs18327	7603 1	594683	С	т	C/T	C/C	C/T	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	
	rs 18754	9062 1	594688 594926	A	G	G/C A/G	G/G A/A	G/C A/A	G/C A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/A	A/A	A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/A	
	rs12445	282 1	594944	С	т	C/C	c/c	c/c	c/c	c/c	c/c	c/c	C/T	c/c	c/c	C/T	c/c	C/T	c/c	c/c	
	rs56342	298 1	595600	С	т	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	
	rs22946	22 1	596259	c	T	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	C/T	C/C	C/C	C/T	C/C	
	rs22356	544 1	597814	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs11864	966 1	597877	С	т	C/T	C/C	C/C	C/C	C/C	c/c	c/c	C/C	C/C	C/C	C/C	C/C	C/C	c/c	c/c	L
	rs22356	45 1	597924	С	т	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	T/T	C/C	C/T	C/T	C/T	C/T	C/C	
	rs 36894	8845 1	599590	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	
	rs22812	28 1	600137	c	Ť	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	т/т	C/C	С/Т	C/T	C/T	C/T	C/C	L
	rs 18921	9366 1	600216	с	G	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	c/c	C/G	C/C	C/C	
	rs 74605	356 1	600272	T	с	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/C	T/T	T/T	T/C	T/T	T/C	T/T	T/T	
	rs22812	29 1	601287	G	A	G/G	G/G A/A	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A G/G	G/G	G/A	G/G	G/G	
	rs22356	647 1	601331	c	A	C/A	C/C	C/C	C/C	C/A	c/c	C/A	c/c	A/A	C/C	C/A	C/A	C/A	C/A	c/c	
	rs26676	64 1	601458	А	G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	
	rs22812	30 1	601767	T	C T	T/C	T/T	T/T	T/T	T/C	T/T	T/C	T/T	C/C	T/T	T/C	T/C	T/C	T/C	T/T	
-	rs80150 rs22813	31 1	601879	A	G	A/G	A/A	A/A	A/A	6/C	C/C A/A	C/C A/G	A/G	G/G	A/A	G/G	A/G	C/C	A/G	C/C A/A	
	rs 79038	3790 1	602035	С	т	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	C/C	C/C	C/C	c/c	C/C	C/C	L
	rs22812	32 1	602114	С	A	C/A	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	
	rs80502	274 1	602260	C	Т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C	1
	rs22812 rs80513	29 1	602323	A	c	A/A	A/A	A/A	A/A	1/1 A/A	A/A	1/1 A/A	A/C	1/1 A/A	A/A	A/C	A/A	A/C	1/1 A/A	1/1 A/A	
	rs80495	66 1	602594	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G	
	rs11409	894 1	602649	С	СТ	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	
	rs55977	7878 1	602649	C	СТ	C/C	C/C	C/C	C/C	C/CT	C/C	C/CT	C/CT	C/CT	C/C	C/C	C/C	C/CT	C/CT	C/C	
	rs11162 rs80518	1420 1	602811	A	G	A/G	A/A	A/A	A/A	A/G	6/C	A/G	A/G	G/G	A/A	G/G	A/G	6/6	A/G	6/C	
	rs80519	09 1	602845	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A	
	rs22812	34 1	602938	А	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A	
	rs22812	35 1	603535	T	c	T/C	T/T	T/T	T/T	T/C	T/T	T/C	T/C	C/C	T/T	C/C	T/C	C/C	T/C	T/T	
	rs22812	075 1	603716	c	A	C/C	C/C	C/C	C/C	G/A C/C	G/G C/C	G/A C/C	G/A C/A	A/A C/C	C/C	C/A	C/C	A/A C/A	G/A C/C	G/G C/C	
	rs66798	3554 1	604388	G	A	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G	1
	rs22729	76 1	604701	T	С	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/C	T/T	T/T	T/C	T/T	T/C	т/т	T/T	L
	-	84 1	605051	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A	
	rs99329		COFOCT		Г	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	1.10	C/T	C/C			CIC	
	rs99325 rs12443	1939 1 61 1	605053 605163	c c	Δ	C/C	C/C	C/C	C/C	C/A	C/C	C/A	C/C	A/A	C/C	C/A	C/A	C/T	C/C	c/c	
	rs99329 rs12443 rs99226 rs11402	1939 10 661 10 22362 10	605053 605163 605234	C T	A C	C/C T/T	C/C T/T	C/C T/T	С/С Т/Т	C/A T/T	C/C T/T	C/A T/T	C/C T/T	A/A T/T	C/C C/C T/T	C/A T/T	C/A T/C	C/T C/A T/T	C/C C/A T/T	C/C C/C T/T	
	rs 99329 rs 12443 rs 99226 rs 11402 rs 10576	1939 10 561 10 12362 10 510 10	605053 605163 605234 605537	C T T	A C C	C/C T/T T/C	C/C T/T T/T	C/C T/T T/T	C/C T/T T/T	C/A T/T T/C	C/C T/T T/T	C/A T/T T/C	C/C T/T T/C	A/A T/T C/C	с/с С/С Т/Т Т/Т	C/A T/T C/C	C/A T/C T/C	C/T C/A T/T C/C	C/C C/A T/T T/C	C/C T/T T/T	
	rs99325 rs12443 rs99226 rs11402 rs11402 rs10576 rs17135	1939 10 161 10 12362 10 100 10 10391 10	605053 605163 605234 605537 605606	C T T C	A C T	C/C T/T T/C C/C	C/C T/T T/T C/C	C/C T/T T/T C/C	C/C T/T T/T C/C	C/A T/T T/C C/T	C/C T/T T/T C/C	C/A T/T T/C C/T	C/C T/T T/C C/C	C/C A/A T/T C/C T/T	C/C T/T T/T C/C	C/A T/T C/C C/T	C/A T/C T/C C/T	C/T C/A T/T C/C C/T	C/C C/A T/T T/C C/T	C/C T/T T/T C/C	
	rs99325 rs12442 rs99226 rs11402 rs10576 rs17133 rs80483	1939 10 1661 10 12362 10 1610 10 16391 10 182 10	605053 605163 605234 605537 605606 605975 606167	C T C C	A C T T	C/C T/T T/C C/C C/T	C/C T/T T/T C/C C/C	C/C T/T T/T C/C C/C	C/C T/T T/T C/C C/C	C/A T/T T/C C/T C/C	C/C T/T T/T C/C C/C	C/A T/T T/C C/T C/C	C/C T/T C/C C/C C/C	C/C A/A T/T C/C T/T C/C	C/C T/T T/T C/C C/C	C/A T/T C/C C/T C/C	C/A T/C T/C C/T C/C	C/T C/A T/T C/C C/T C/C	C/C C/A T/T T/C C/T C/C	C/C T/T T/T C/C C/C	
	rs99325 rs12442 rs99226 rs11402 rs10576 rs17733 rs80483 rs80533 rs80533 rs18005	1939 1 1661 1 12362 1 10 1 1391 1 182 1 135 1 10290 1	605053 605163 605234 605537 605606 605975 606167 606336	C T C C A C	A C T G T	C/C T/T T/C C/C C/T A/G C/C	C/C T/T T/T C/C C/C A/A C/C	C/C T/T C/C C/C A/A C/C	C/C T/T T/T C/C C/C A/A C/C	C/A T/T C/C C/T C/C A/A C/C	C/C T/T C/C C/C A/A C/C	C/A T/T T/C C/T C/C A/A C/C	C/C T/T C/C C/C C/C A/A C/C	C/C A/A T/T C/C T/T C/C A/A C/C	C/C T/T T/T C/C C/C A/A C/C	C/A T/T C/C C/T C/C A/A C/C	C/A T/C T/C C/T C/C A/A C/C	C/T C/A T/T C/C C/T C/C A/A C/C	C/C C/A T/T C/T C/T C/C A/A C/C	C/C T/T T/T C/C C/C A/A C/C	

2																				
3 1 1 5 1 5 1 6 5 5 5 5 5 6	2																			
	3	rs138001290	1606336	C	TG	C/C A/G	C/C A/A	C/C A/A	C/C A/A	C/C	C/C	C/C	C/C	C/C	C/C A/A	C/C G/G	C/C A/G	C/C	C/C	C/C
4 production 1 0	5	rs12599207	1606445	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A
5 minima	4	rs55963259 rs12927247	1606479 1606739	C A	G	C/T A/A	C/C A/A	C/C A/A	C/C A/A	C/C A/G	C/C A/A	C/C A/G	C/C A/A	C/C G/G	C/C A/A	C/C A/G	C/C A/G	C/C A/G	C/C A/G	C/C A/A
	5	rs112810668	1607113	GC	G	GC/G	GC/GC	GC/GC	GC/GC	GC/GC	GC/GC	GC/GC	GC/G	GC/GC	GC/GC	GC/G	GC/GC	GC/G	GC/GC	GC/GC
7 8 8 8 8 8 8 9 10	6	rs116905776 rs11642907	1607242	G	T	G/G	G/G	G/G	G/G	G/G	G/G	G/G	C/C G/T	G/G	G/G	G/T	G/G	G/T	G/G	G/G
6 1	7	rs78753725	1607286	G	C C	G/C	G/G	G/G	G/G T/T	G/G	G/G T/T	G/G	G/G	G/G T/T	G/G T/T	G/G	G/G T/T	G/G	G/G	G/G
8 abox bbox bbox <t< th=""><th>/</th><th>rs2076435</th><th>1607574</th><th>c</th><th>т</th><th>C/T</th><th>c/c</th><th>c/c</th><th>c/c</th><th>C/T</th><th>c/c</th><th>C/T</th><th>с/т</th><th>т/т</th><th>c/c</th><th>T/T</th><th>C/T</th><th>т/т</th><th>C/T</th><th>C/C</th></t<>	/	rs2076435	1607574	c	т	C/T	c/c	c/c	c/c	C/T	c/c	C/T	с/т	т/т	c/c	T/T	C/T	т/т	C/T	C/C
9 9 1 10 10 10 10 10 10 10 10 10 10 10 10 1	8	rs2076436 rs2235641	1608082 1608177	A	G	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/G A/G	A/A A/A	A/G A/G	A/A A/G	G/G G/G	A/A A/A	A/G G/G	A/G A/G	A/G G/G	A/G A/G	A/A A/A
	9	rs12446290	1608364	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
	10	rs 74821318 rs 540739250	1608729 1608904	c	A T	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/A C/C	G/G C/C	G/G C/C	G/A C/C	G/G C/C	G/A C/T	G/G C/C	G/G C/C
11 Image: 10 into 1 / 10 into 1	10	rs56289115	1608934	A	G	A/G	A/A T/T	A/A T/T	A/A T/T	A/G	A/A	A/G	A/G	G/G	A/A T/T	G/G	A/G	G/G	A/G	A/A
12	11	rs145464572	1609624	c	A	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
13 Image: Note of the set of th	12	rs3784835 rs34316191	1609843 1609944	G	С	G/G C/C	G/G C/T	G/G C/T	G/G C/T	G/G	G/G	G/G	G/C	G/G	G/G C/C	G/C	G/G C/C	G/C	G/G	G/G
144 135 1	13	rs3784836	1610176	G	Å	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
144 Number	14	rs 3784837 rs 2003343	1610215 1610578	G	G	C/G G/G	C/C G/G	C/C G/G	C/C G/G	C/C G/G	C/C G/G	C/C G/G	C/G G/G	C/C G/G	C/C G/G	C/G G/G	C/C G/G	C/G G/G	C/C G/G	C/C G/A
15 Image: Balance Balanc	14	rs1016499	1610885	A	G	A/G	A/A	A/A	A/A	A/A	A/A	A/A	A/G	A/A	A/A	A/G	A/A	A/G	A/A	A/A
16 Image: Base of the set of th	15	rs188815635	1612145	C	G T	C/C	C/C	C/C	C/C	C/C	GI/G C/C	C/C	GI/G C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
17 Number 1 c </th <th>16</th> <th>rs3784838</th> <th>1612498</th> <th>C A</th> <th>G</th> <th>C/G</th> <th>C/C</th>	16	rs3784838	1612498	C A	G	C/G	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
1/1 1/2 0 1/2	17	rs12446060	1613365	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
18 Treeded 14.000 C A <	17	rs184721812 rs28714146	1613428 1613793	T A	C C	T/T A/A	T/T A/A	T/T A/A	T/T A/A	T/T A/C	T/T A/A	T/T A/C	T/T A/A	T/T C/C	T/T A/A	T/T A/C	T/T C/C	T/T A/C	T/T A/C	T/T A/A
19 10 <th< th=""><th>18</th><th>rs778960409</th><th>1614093</th><th>G</th><th>A</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th><th>G/G</th></th<>	18	rs778960409	1614093	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
20 ======= ===== ===== ==== ==== ==== ==== == = <th>19</th> <th>rs8048410 rs11248883</th> <th>1614097 1614240</th> <th>A C</th> <th>G</th> <th>A/G C/C</th> <th>A/A C/C</th> <th>A/A C/C</th> <th>A/A C/C</th> <th>A/G C/C</th> <th>A/A C/C</th> <th>A/G C/C</th> <th>A/G C/G</th> <th>G/G C/C</th> <th>A/A C/C</th> <th>G/G C/G</th> <th>G/G C/C</th> <th>G/G C/G</th> <th>A/G C/C</th> <th>A/A C/C</th>	19	rs8048410 rs11248883	1614097 1614240	A C	G	A/G C/C	A/A C/C	A/A C/C	A/A C/C	A/G C/C	A/A C/C	A/G C/C	A/G C/G	G/G C/C	A/A C/C	G/G C/G	G/G C/C	G/G C/G	A/G C/C	A/A C/C
21 1	20	rs17135407	1615189	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
21	20	rs11648609	1616528	т	A	с/с т/т	T/T	T/T	T/T	C/C	c/c	c/c	C/T	c/c	T/A	T/T	T/T	C/T	c/c	c/c
223 1	21	rs11648819 rs8059442	1616663	C	T	C/C	C/C G/G	C/C G/G	C/C G/G	C/C	C/C	C/C	C/C	C/C	C/C G/G	C/T G/G	C/C G/G	C/C	C/C	C/C
23	22	rs77486198	1616838	c	т	c/c	c/c	c/c	c/c	c/c	C/C	c/c	C/T	c/c	c/c	C/T	C/C	C/T	c/c	C/C
24 1 1 2 4	23	rs7186358 rs11641175	1617688 1618007	C C	G	C/G C/C	C/C C/C	C/C C/C	C/C C/C	C/C C/C	c/c c/c	c/c c/c	C/G C/A	c/c c/c	C/C C/C	C/G C/A	C/C C/C	C/G C/A	c/c c/c	c/c c/c
24-4	24	rs11646462	1618112	A	С	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/C	A/A	A/A	A/C	A/A	A/C	A/A	A/A
25 memory and a set of a base	24	rs180759409	1618995	c	т	C/T	C/C	C/T	C/T	C/C	C/C	c/c	C/C	c/c	C/C	C/C	C/C	C/C	c/c	C/C
26 192721 193841 C 1 or ccc cc ccc cccc ccc ccc c	25	rs9927187	1619074	C T	T C	C/T T/C	C/C T/T	C/C T/T	C/C T/T	C/C T/T	C/C T/T	C/C T/T	C/C	C/C T/T	C/C T/T	C/T T/C	C/C T/T	C/T T/C	C/C T/T	C/C T/T
27 mm32733 Bisses C T Gr	26	rs9927511	1619431	c	т	С/т	c/c	c/c	c/c	c/c	c/c	c/c	с/т	c/c	c/c	с/т	C/C	C/T	c/c	c/c
11180211 1118078	27	rs9927529 rs181102139	1619488 1619703	C T	T A	C/T T/T	C/C T/T	C/C T/T	C/C T/T	C/T T/T	C/C T/T	C/T T/T	C/T T/T	T/T T/T	C/C T/T	T/T T/T	C/T T/T	T/T T/T	C/C T/T	C/C T/T
260	20	rs113900211	1619758						ITTTT/ATTT											TTTT/ATTTT
299 null201189 hul20189 t	28	rs551270839	1619758	A	AT,ATT	A/AI A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/A	A/A A/ATT	A/A A/AT	A/A A/A	A/A A/A	A/A A/A	A/A A/ATT	A/A A/A	A/A A/A
300 1938888 1938888 1938888 193888 193888 193888 1938888 1938888 1938888 1938888 1938888 1938888 1938888 1938888 1938888 19388888 1938888888 1938888888888 19388888888888 1938888888888888 193888888888888888 193888888888888888888888888888888888 1938888888888888888	29	rs117401189	1619761	T	G	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	т/т			T/T	T/T
31 ph/499705 123097 C T C	30	rs7200291	1619896		C	0/0	0/0	0,0	0/0		0/0	0/0			6/6	6/6	T/G	T/T G/G	6/6	G/G
1 n	31			A	т	A/T	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	G/G A/A	G/G A/A	T/G G/G A/A	T/T G/G A/A	G/G A/A	G/G A/A
32 1/2/2011 100/00 1 1 100/00 1	JI	rs118102263	1620307 1620424	C G	T T C	A/T C/C G/G	A/A C/T G/G	A/A C/C G/G	A/A C/C G/G	A/A C/C G/G	A/A C/C G/G	A/A C/C G/G	A/A C/C G/G	6/G A/A C/C G/G	G/G A/A C/C G/G	G/G A/A C/C G/G	T/G G/G A/A C/C G/C	T/T G/G A/A C/C G/G	G/G A/A C/C G/G	G/G A/A C/C G/G
333 (19986297) c) c)	22	rs149487053 rs118102263 rs76069945	1620307 1620424 1620468	G G	T C C	A/T C/C G/G G/G	A/A C/T G/G G/G	A/A C/C G/G G/G	A/A C/C G/G G/G	A/A C/C G/G G/G	A/A C/C G/G G/G	A/A C/C G/G G/G	6/ G A/A C/C G/G G/C	G/G A/A C/C G/G G/G	G/G A/A C/C G/G G/G	G/G A/A C/C G/G G/C	T/G G/G A/A C/C G/C G/G	T/T G/G A/A C/C G/G G/C	G/G A/A C/C G/G G/G	G/G A/A C/C G/G G/G
34 19393322 193292 1	32	rs 144487053 rs 118102263 rs 76069945 rs 72761116 rs 12448197	1620307 1620424 1620468 1620550 1621119	G G G G	T C C T C	A/T C/C G/G G/G C/T G/G	A/A C/T G/G G/G C/C G/G	A/A C/C G/G G/G C/C G/G	A/A C/C G/G G/G C/C G/G	A/A C/C G/G G/G C/C G/G	A/A C/C G/G G/G C/C G/G	A/A C/C G/G G/G C/C G/G	A/A C/C G/G G/C C/C G/C	G/G A/A C/C G/G C/C G/G	G/G A/A C/C G/G G/G C/C G/G	G/G A/A C/C G/G G/C C/C G/C	T/G G/G A/A C/C G/C G/G C/C G/G	T/T G/G A/A C/C G/G G/C C/C G/C	G/G A/A C/C G/G G/G C/C G/G	G/G A/A C/C G/G G/G C/C G/G
1 1	32 33	rs 14948/053 rs 118102263 rs 75069945 rs 72761116 rs 12448197 rs 199826737 rs 72481027	1620307 1620424 1620468 1620550 1621119 1621495 1621702	A G G C C C	T C C T C T	A/T C/C G/G G/G C/T G/G C/C	A/A C/T G/G G/G C/C G/G C/C	A/A C/C G/G G/G C/C G/G C/C	A/A C/C G/G G/G C/C G/G C/C	A/A C/C G/G G/G C/C G/G C/C	A/A C/C G/G G/G C/C G/G C/T	A/A C/C G/G C/C C/C C/C	A/A C/C G/G G/C C/C G/C C/T	G/G A/A C/C G/G G/G C/C G/G C/C	G/G A/A C/C G/G G/G C/C C/C	G/G A/A C/C G/G G/C C/C G/C C/C	T/G G/G A/A C/C G/C G/G C/C G/G C/C	T/T G/G A/A C/C G/G G/C C/C G/C C/C	G/G A/A C/C G/G G/G C/C G/G C/C	G/G A/A C/C G/G G/G C/C C/C C/C
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34	rs 118102263 rs 76069945 rs 76069945 rs 72761116 rs 12448197 rs 79826737 rs 72481037 rs 72481037	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967	G G C C C T	т с с т с т с	A/T C/C G/G G/G C/T G/G C/C C/C C/C T/T	A/A C/T G/G G/G C/C G/G C/C C/C C/C T/T	A/A C/C G/G G/G C/C G/G C/C C/C T/T	A/A C/C G/G G/G C/C C/C C/C C/C T/T	A/A C/C G/G G/G C/C G/G C/C C/C C/C T/T	A/A C/C G/G C/C C/C C/T C/C T/T	A/A C/C G/G C/C G/G C/C C/C C/C T/T	A/A C/C G/G G/C C/C C/T C/T T/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C T/T	G/G A/A C/C G/G G/G C/C C/C C/C C/C T/T	G/G A/A C/C G/G G/C C/C C/C C/T T/C	T/G G/G A/A C/C G/C G/G C/C C/C C/C T/T	T/T G/G A/A C/C G/G G/C C/C G/C C/C C/T T/C	G/G A/A C/C G/G G/G C/C G/G C/C C/C T/T	G/G A/A C/C G/G G/G C/C C/C C/C C/C T/T
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34	15.14946/033 15.118102263 15.726069945 15.72761116 15.12448197 15.194826737 15.7248107 15.7248107 15.79963862 15.17135411 15.58856196	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967 1622008 1622092	G G C C C T A C	т с с т с т с с т	A/T C/C G/G G/G C/T G/G C/C C/C C/C T/T A/G C/T	A/A C/T G/G G/G C/C G/G C/C C/C C/C T/T A/A C/C	A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	A/A C/C G/G G/G C/C C/C C/C C/C T/T A/A C/C	A/A C/C G/G C/C G/G C/T C/C T/T A/A C/C	A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C	A/A C/C G/G C/C C/C C/T C/T T/C A/G C/T	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C A/G C/T	T/G G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	T/T G/G A/A C/C G/G C/C C/C C/C C/T T/C A/G C/T	7/1 G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C	G/G A/A C/C G/G G/G C/C G/G C/C C/C C/C T/T A/A C/C
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34 35	15.4948/033 rs118102263 rs72060945 rs7276116 rs12448197 rs1294826737 rs7281077 rs59963862 rs17135411 rs58856196 rs58334533	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967 1622008 1622092 1622092	A C G G C C C T A C G	T C C T C T C G T A	A/T C/C G/G G/G C/T G/G C/C C/C C/C T/T A/G C/T G/A	A/A C/T G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G	A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G	A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G	A/A C/C G/G C/C C/C C/C C/C C/C T/T A/A C/C G/G	A/A C/C G/G C/C G/G C/T C/C T/T A/A C/C G/G	A/A C/C G/G C/C C/C C/C C/C T/T A/A C/C G/G	A/A C/C G/G C/C C/C C/T C/T T/C A/G C/T G/A	G/G A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G	G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C A/G C/T G/A	T/G G/G A/A C/C G/C G/G C/C C/C C/C T/T A/A C/C G/G	T/T G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C A/G C/T G/A	7/1 G/G A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C T/T A/A C/C G/G
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34 35 36	15 14940/03 15 118102263 15 7000945 15 72761116 15 12448197 15 199826737 15 7248107 15 79963862 15 71735411 15 7589365196 15 758334533 15 190238492 15 7022565	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967 1622008 1622092 1622108 1622208 16222539 1623251	A C G G C C C C C C C C C C G C G C G C	T C C T C T C G T A T A	A/T C/C G/G G/G C/T G/G C/C C/C C/C T/T A/G G/A C/C G/A	A/A C/T G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G	A/A C/C G/G C/C G/G C/C C/C C/C T/T A/A C/C G/G C/T G/G	A/A C/C G/G C/C C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G	A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/C G/G	A/A C/C G/G C/C G/G C/T C/C T/T A/A C/C G/G C/T G/G	A/A C/C G/G C/C G/G C/C C/C T/T A/A C/C G/G C/T G/G	A/A C/C G/G G/C C/C C/T C/T T/C A/G C/T G/A C/C G/A	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/C G/C	G/G A/A C/C G/G C/C C/C C/C T/T A/A C/C G/G C/C G/G	G/G G/A G/G G/C C/C C/C C/C C/T T/C A/G C/T G/A C/C G/A	T/G G/G A/A C/C G/G C/C C/C C/C C/C T/T A/A C/C G/G G/G	T/T G/G A/A C/C G/G C/C C/C C/C C/C C/C T/C A/G C/T G/A C/C G/A	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C G/G C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34 35 36 37	15.14948/053 15.116102263 15.72761116 15.12448197 15.19826737 15.2986382 15.17135411 15.58856196 15.83834533 15.190238492 15.722555 15.127940418 15.722655 15.127940418 15.7260132	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967 1622008 1622092 162209 162209 1622539 1623251 1623473 1623473	A G G C C C T A C G C TIGTAT	T C C T C T C G T A T A T C	A/T C/C G/G G/G C/T G/G C/C C/C C/C G/A C/C G/A C/C G/A C/C C/C	A/A C/T G/G G/G G/G C/C C/C C/C T/T A/A C/C G/G C/T G/G STATT/CTTG	A/A C/C G/G G/G C/C C/C C/C C/C C/T A/A C/C G/G G/G STATT/CTTG	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/T G/G G/G C/T G/G G/T	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C G/G C/C C/C	A/A C/C G/G C/C C/C C/C T/T A/A C/C C/C C/T G/G STATT/CTTG	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/C C/C C/T C/T T/C A/G C/T G/A C/T G/A C/C G/A	6,6 A/A C/C G/G G/G C/C C/C T/T A/A C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/C G/G STATT/CTTG	G/G G/A A/A C/C G/C C/C C/C C/C C/T T/C A/G C/T G/A C/C G/A C/C G/AT//	T/G G/G A/A C/C G/G C/C C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C G/G C/C	T/T G/G A/A C/C G/G C/C C/C C/C C/C C/T T/C A/G C/T G/A C/C G/A C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C
$ \begin{array}{c ccccccccccccccccccccccccccccccccccc$	32 33 34 35 36 37 38	1514948/053 ri116102263 rs72761116 rs12448197 rs199826737 rs2986382 rs17135411 rs88856196 rs83834533 rs190238492 rs722555 rs127394018 rs77960132 rs13380687	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1621967 1622008 1622092 1622208 1622539 1623251 1623473 1623638 1623852	A G G C C C T A C G C TIGTAT C A	T C C T C T C G T A T C G T G	A/T C/C G/G G/G C/T G/G C/C T/T A/G C/T G/A C/T G/A CTIGTATT/C C/C A/G	A/A C/T G/G G/G C/C G/G C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G G/A C/T C/T G/G C/C C/C C/C	A/A C/C G/G C/C C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G C/C C/T C/C T/T C/C T/T A/A C/C G/G C/T G/G STATT/CTTG C/C A/A	A/A C/C G/G G/G C/C C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G C/T G/G C/C C/A/A	A/A C/C G/G G/C C/C C/T C/T T/C A/G C/T G/A C/C G/A C/C G/A C/C A/G	6,06 A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C C/C	G/G A/A C/C G/G G/C C/C C/C C/T T/C A/G C/T G/A C/C G/A C/C G/A C/C G/A	T/G G/G A/A C/C G/G C/C G/G C/C C/C C/C C/C G/G C/C G/G C/C C/C	T/T G/G A/A C/C G/G G/C C/C C/T T/C A/G C/T G/A C/C G/A C/C G/A C/C G/A	, , , , , , , , , , , , , , , , , , ,	G/G A/A G/G G/G G/G C/C C/C C/C C/C C/C C/C G/G C/T G/G C/T G/G C/T G/G
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34 35 36 37 38	15 14948/053 ri116102263 rs72761116 rs72761116 rs1294826737 rs72481037 rs744413 rs77960132 rs744413 rs744414 rs744414 rs744414 rs74441	1620307 1620424 1620468 1620550 1621119 1621495 1621702 1622008 1622092 1622008 1622208 1622208 1622539 1623251 1623473 1623638 1623852 1624406	A G G C C C T A C G C T G C T G T T	T C C T C T T C C T T C C C C C C C C C C C C C C C C C C C C	A/T C/C G/G G/G C/T C/C C/C T/T A/G C/T G/A C/T C/C C/C C/C C/C C/C C/C C/C	A/A C/T G/G G/G C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/C G/G C/C G/G C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/T G/G STATT/CTTG C/C A/A C/C C/T T/T	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C G/G C/C C/C	A/A C/C G/G C/C C/T C/C T/T A/A C/C G/G C/T G/G ETATT/CTTG C/C A/A T/T	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A A/A C/C G/C C/C C/C C/T T/C A/G C/T G/A C/C C/C G/A C/C C/C C/T T/C A/G T/T T/C	6)(6) A/A C/C G/G G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/C G/G C/C C/C A/A T/T	G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C A/G C/T G/A C/C G/A C/C G/A C/C C/C G/A C/C C/C G/A C/C C/C T/T T/C	T/G G/G A/A C/C G/G C/C G/G C/C C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	T/T G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C G/A C/T G/A C/T G/A C/C G/A C/C C/C G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	, , , , , , , , , , , , , , , , , , ,	G/G A/A G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C
$ \begin{array}{c c c c c c c c c c c c c c c c c c c $	32 33 34 35 36 37 38 39	1514948/053 rs118102263 rs72761116 rs122448197 rs199826737 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs744103413 rs78343453 rs79760132 rs72480687 rs147960132 rs13380687 rs147960132 rs12396975 rs1479601463 rs1090001 rs12596975 rs1090001 rs12596975 rs1090001 rs12596975 rs1090001 rs12596975 rs1090001 rs12596975 rs1090001 rs12596975 rs10900001 rs12596975 rs10900000 rs12596975 rs10900000000000000000000000000000000000	1620307 1620424 1620458 1620550 1621119 1621495 1621495 1622092 1622092 162208 1622092 1622108 162251 162351 1623473 162352 162352 1623473	A G G C C C T A C C T G C T T T G	T T C C T C T T C G T A T A C T G C C A T	A/T C/C G/G G/G C/T C/C C/C T/T G/A C/C G/A C/C C/C A/G T/C C/C C/C C/C C/C C/C C/T C/C C/C	A/A C/T G/G G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G G/G	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G G/G	A/A C/C G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/C G/G C/C G/G C/C A/A T/T T/T T/T G/G	A/A C/C G/G G/G C/C C/T C/C T/T A/A C/C G/G T/T T/T T/T T/T G/G C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A A/A C/C G/C G/C C/C C/T T/C A/G C/T G/A C/C C/C A/G T/T T/C G/A C/C C/C A/G T/T	6,6 4,7 4,7 6,7 6,7 6,7 6,7 6,7 6,7 6,7 6,7 6,7 6	G/G A/A C/C G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G G/G C/C A/A T/T T/T G/G C/C	G/G A/A C/C G/G G/C C/C C/C C/T T/C G/A C/C G/A C/C G/A C/C G/A C/C C/C C/C C/C C/T T/C G/A C/C	7/G G/G A/A C/C G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G STATT/CTTC C/C A/A T/T T/T T/T	T/T G/G A/A C/C G/G G/C C/C C/C C/C C/C C/C C/T T/C G/A C/T G/A C/C C/C G/A C/C C/C G/A C/C C/C G/A C/C G/A A G/C C/C C C/C C C/C C C/C C C C C C C C	, , , , , , , , , , , , , , , , , , ,	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C
$\begin{array}{cccccccccccccccccccccccccccccccccccc$	32 33 34 35 36 37 38 39 40	15.4948/033 ri116102263 rs72761116 rs12248197 rs199826737 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs72481037 rs74410341 rs783430431 rs7960132 rs132340418 rs79760132 rs132340687 rs147054463 rs12596975 rs112369751 rs13230451	1620307 1620424 1620468 1620468 162050 162119 162119 162197 1622008 1622008 1622008 1622008 16220208 16220208 1622408 1622473 1623473 1623452 1624403 1624852 1624493 1624493	A G G C C C T A C C G C C T G C C T G T T T G C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С	A/T C/C G/G G/G G/G G/G G/C C/C C/C	A/A G/G G/G G/G C/C C/C C/C T/T A/A C/C G/G C/T G/G C/C C/C A/A T/T T/T G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G G/G C/C C/C C/C C/C	A/A C/C G/G G/G G/G C/C C/C C/C C/T A/A C/C G/G C/T C/T C/T C/T C/T C/T C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/T C/C T/T A/A C/C G/G C/T G/G C/C A/A T/T T/T G/G C/C C CCCT/CCCCC	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/C G/C C/T C/T T/C C/T C/T G/A C/C G/A C/C G/A C/C C/C A/G T/T T/C G/A C/C C/C C/T C/T C/T C/T C/T C/T	6) 50 A/A C/C G/G C/C C/C C/C C/C C/C C/C	6/G A/A C/C G/G G/G C/C C/C C/C C/C C/C G/G C/C G/G C/C A/A T/T T/T G/G C/C C/C CCCT/CCCC	G/G A/A C/C G/G G/C C/C C/C C/T T/C G/A C/C G/A C/C G/A C/C C/C C/C C/T T/C G/A C/C C/C C/C C/C C/C C/C C/C C/C C/C C	T/G G/G A/A C/C G/G C/C C/C C/C C/C C/C G/G C/C G/G C/C C/C	T/T G/G A/A C/C G/G G/G G/C G/C G/C G/C G/C G/C G/G G/A	Г/Т G/G A/A C/C G/G G/G G/G C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C
42 fr:11192403 1520059 G G	32 33 34 35 36 37 38 39 40 41	15 14948/053 15 118102263 15 72761116 15 12448197 15 199826737 15 72481037 15 72481037 15 72481037 15 735411 15 85856196 15 735451 15 735555 15 735555 15 7355555 15 7355555 15 7355555 15 7355555 15 7355555 15 73555555 15 7355555555555555555555555555555555555	1620307 1620424 1620468 1620468 162050 162119 162119 162119 162109 162109 162109 1622092 1622092 1622092 1622092 1622208 1622351 162249 162249 162249 162249 162249 162442 16245 16255 16255 16255 16255 16255 162555 162555 162555 162555 1625555 16255555 16255555 1625555555555	A G G C C C C T A C G C C C C C C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G	А/Т С/С G/G G/G G/G G/G C/C C/C C/C C/C	A/A G/G G/G G/G C/C C/C C/C T/T A/A C/C G/G C/C G/G C/C C/C T/T T/T T/T G/G C/C C/C A/A C/C C/C C/C C/C C/C	А/А с/С G/G G/G G/G C/C C/C C/C C/C C/C	Α/Α C/C G/G G/G G/G C/C C/C C/C C/T A/A C/C G/G C/T T/T G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/T C/C C/C T/T A/A C/C G/G C/T G/G C/T T/T T/T G/G C/C A/A T/T T/T G/G C/C A/A	A/A C/C G/G G/G C/C C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G C/T T/T T/T T/T T/T T/T T/T T/T	A/A C/C G/C G/C C/T C/T T/C A/G C/T C/T G/A C/C G/A C/C G/A C/C C/T T/C G/A C/C C/T T/C C/T C/T	6),6 A/A C/C G/G C/C C/C C/C C/C C/C C/C	6/6 A/A C/C 6/6 6/6 C/C C/C C/C C/C T/T A/A C/C G/6 C/C G/6 C/C C/C A/A T/T T/T G/6 C/C C/C C/C T/T T/T	G/G A/A C/C G/G G/C C/C C/C C/T T/C G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C C C/C C/C C/T T/C G/A C/C C C/C C/C C/C C/C C/C C/C C/C C/C	T/G G/G A/A C/C G/G G/G C/C G/G C/C C/C C/C C/C C/C G/G G/G JAPA C/C G/G G/G G/G C/C G/G C/C G/G C/C A/A A/A T/T T/T G/G C/C C/C C/C C/C C/C C/C C/C T/T T/G C/C C/CCCT/CCCI	T/T G/G G/A C/C G/G G/G G/C G/C G/C G/C G/C G/C G/C G/A C/T G/A C/C G/A	Г/Г G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C
43 s326207 1620227 T TA T/TA T/T	32 33 34 35 36 37 38 39 40 41	15 14948/053 15 118102263 15 72761116 15 12448197 15 199826737 15 7246137 15 72761136 15 72761136 15 73364137 15 735411 15 85856196 15 85834533 15 190238492 15 727960132 15 132300687 15 137940418 15 132300687 15 137940418 15 12596975 15 112369751 15 138310551 15 30045127 15 13830456	1620307 1620424 1620468 1620468 162050 162119 162119 162119 1621967 1622092 1622092 1622092 1622092 1622208 1622351 162363 1623638 1623632 162442 16244288 16244288 1624784 1624784	A G G C C C C T T C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С	А/Т С/С G/G G/G C/T G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/T G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/C C/T G/G C/C C/T T/T G/G C/C C/C T/T T/T T/T	Α/Α C/C G/G G/G G/G C/C C/C T/T Α/Α C/C G/G C/T G/G C/T C/C C/T C/C C/T T/T T/T G/G C/C C/C T/T T/T	А/А G/G G/G G/G C/C G/G C/C C/C T/T A/A C/C G/G C/C G/G C/C C/C T/T T/T G/G C/C T/T T/T	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/T C/C T/T A/A C/C G/G C/T G/G C/T G/G C/T T/T T/T G/G C/C T/T T/T T/T	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/C G/C C/T C/T C/T G/A C/C G/A C/C G/A C/C G/A C/C C/T G/A C/C C/T C/T C/T C/T C/T C/T C/T	6) 56 A/A C/C G/G C/C C/C C/C C/C C/C C/C	6/6 A/A C/C G/6 G/6 C/C C/C C/C T/T A/A C/C G/6 C/C G/6 C/C G/6 C/C G/6 C/C C/C T/T T/T T/T T/T T/T	G/G A/A C/C G/G G/C C/C C/T T/C A/G C/T G/A C/C G/A C/T G/A C/C G/A T/T T/C A/G T/T T/C C/C C/T C/C C/T C/C C/C C/C C/C	T/G G/G A/A C/C G/G G/G C/C G/G C/C C/C C/C G/G G/G C/C G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	T/T G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C G/A C/C C/T G/A C/C C/T G/A C/C C/T G/A C/C C/T G/A C/C C C/T T/T T/C G/A C/C C C/C C/C C/C C/C C/C C/C C/C C/C	Г/Г G/G A/A C/C G/G G/G G/G C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C
$\begin{array}{c c c c c c c c c c c c c c c c c c c $	32 33 34 35 36 37 38 39 40 41 42	s 14948/033 si 118102263 s 72761116 s 12248197 s 72761116 s 72761116 s 72761116 s 7276117 s 73481037 s 73481037 s 73481037 s 73481037 s 73481037 s 73481037 s 73481037 s 73481037 s 73490312 s 737960132 s 7379601	1620307 1620424 1620468 1620550 162119 162119 1621495 1621967 1621008 1621967 1622008 162208 1622351 1622351 1622351 1622351 1623453 1624528 162442 162442 162442 1624724 1624724	A G G C C T A C C C C C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G	A/T C/C G/G G/G G/G C/T G/G C/T C/C C/T G/G C/T G/G C/T G/A C/C G/A C/C G/A C/C C/A G/A C/C T/C G/A C/C T/T T/C G/A C/CCCT/C T/T T/C G/A A/G	A/A C/T G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/T G/G C/T C/C C/T C/C C/C T/T T/T G/G C/C C/C T/T T/T G/G A/A	Α/Α C/C G/G G/G G/G C/C C/C C/C C/C G/G C/C G/G C/C G/G C/T G/G C/T G/G T/T T/T G/G C/C C/C C/T G/G T/T T/T T/T G/G CCCCT/CCCCC T/T G/A	А/А С/С G/G G/G C/C C/C C/C C/C T/T A/A C/C G/G C/C G/G C/C C/C T/T T/T G/G C/C T/T T/T G/C C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G G/G C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/C G/C C/T C/T G/A C/T G/A C/C G/A C/C G/A C/C C/T C/T G/A C/C C/T C/T C/T C/T C/T C/T C/T	9,93 A/A C/C G/G G/G C/C C/C C/C C/C C/C	G/G A/A C/C G/G G/G C/C G/G C/C C/C T/T A/A C/C G/G C/C C/C G/G C/C C/C A/A T/T T/T G/G C/C C/C T/T T/T T/T T/T G/G G/G C/C C C/C A/A A A A A A A A A A A A A A	G/G A/A C/C G/G G/C C/C C/T T/C A/G C/T G/A C/T G/A C/C G/A C/T G/A C/C C/T C/C C/T C/C C/T C/C C/T C/C C/T C/C C/T C/C C/C	T/G G/G A/A C/C G/G G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G G/G G/G G/G T/T T/T G/G C/C C/C G/G T/T G/G	T/T G/G A/A C/C G/G G/C C/C C/C C/C C/T T/C G/A C/C C/T T/C G/A C/C C/T T/T T/C G/A C/C C C/T T/G T/G T/G T/G T/G C/C C C/C C/T C C/C C/C C/C C/C C/C C/C	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C C/C G/G C/T G/G C/C G/G C/C C/C G/G C/C C/C C/C G/G C/C C/C C/C C/C G/G T/T T/T T/T T/T G/G C/C C/C
45 is 59783459 1626607 c c A c/c	32 33 34 35 36 37 38 39 40 41 42 43	15.14948/053 rs116102263 rs72761116 rs12448197 rs92826737 rs7276116 rs129826737 rs92963862 rs17135411 rs83835433 rs190238422 rs722555 rs1373900418 rs77960132 rs12938975 rs147054463 rs1296975 rs12369751 rs1330056 rs13330456 rs112380456 rs1330456 rs1348045127 rs1330456 rs13480456 rs13480456 rs13480456 rs13480456 rs13480456 rs13480456 rs13480456 rs1443870 rs1443870 rs1443870 rs12443870 rs1252072	1620307 1620424 1620458 1620458 1620459 162119 162119 162119 1621967 162109 162209 162208 162208 162251 1622473 1622473 1622473 1624528 162473 1624528 1624771 1624528 1624771 1624528	A G G C C C T T C A C C T G C C C T T T G C C C C T T T G C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G ТА Т	A/T C/C G/G G/G C/T G/G C/C C/C C/C C/C C/C C/C G/A C/C G/A C/C C/C C/C C/C C/C C/C C/C C	A/A C/T G/G G/G C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C T/T A/A C/C C/C C/C C/C C/C C/C C/C	А/А С/С G/G G/G C/C G/G C/C C/C C/C C/C	A/A C/C G/G G/G G/G C/C C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	A/A C/C G/G G/G G/G C/T C/C C/C G/G C/T A/A C/C G/G C/T G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/G G/C C/T T/C G/A C/C G/A C/C C/C A/G G/A C/C C/C A/G C/C C/C C/T T/C G/A C/C C/C C/C C/T T/C G/A C/C C/T T/C C/T T/C C/T T/C C/C C	6) 69 A/A C/C G/G G/G G/G C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	G/G A/A C/C G/C G/C C/C C/C C/C C/C C/C C/C C/C	Τίδα Giβ G/G A/A A/A C/C G/G G/C G/C G/C G/C C/C T/T G/G G/G G/G T/T G/G T/G T/G T/G T/G T/G T/G T/G T/G T/G T/G G/G A/A	ТТ G/G A/A C/C G/G G/G G/C C/C G/G C/C C/C C/C C/C C/C C/C C/C G/A G/A C/C G/A C/C	Г/Т G/G A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	G/G A/A A/A G/G G/G G/G G/G C/C C/C C/C C/C C/C C/C
$ \begin{array}{c} 1 \\ 5 \\ 1 \\ 1 \\ 1 \\ 1 \\ 1 \\ 1 \\ 1 \\ 1 \\$	32 33 34 35 36 37 38 39 40 41 42 43 44	15.14948/063 rs116102263 rs72761116 rs12448197 rs19826737 rs59963862 rs17335411 rs8834353 rs199226737 rs59963862 rs17335411 rs8834533 rs190238422 rs722555 rs137390418 rs77960132 rs13300687 rs147054463 rs12596975 rs1330051 rs1330051 rs12369751 rs1330056 rs112269751 rs13300456 rs11294003 rs1129403 rs12526272 rs12443870	1620307 1620424 1620458 1620468 1620459 1621109 1621195 1621109 16211957 1622008 1622008 1622019 16222108 1622351 1623451 1623451 1623452 1624528 1624771 1624528 1624771 1624528 1624771 1624528 1624771 1624528 1624771 1625890 1626058 1626058 1626058	A G G C C C T T C C T G G C T G T T T G G C C C T T T T	T T C C T T C G G T A T A C T G C C A T C G G C A G TA C C A	A/T C/C G/G G/G C/T G/G C/C C/C T/T A/G C/C G/A C/C G/A C/C C/C C/C C/C T/T T/C G/A C/T C/T T/T C/C C/T C/T C/T C/T	А/А С/Т G/G G/G C/C C/C C/C C/C C/C C/C	A/A G/G G/G G/G C/C G/G C/C C/C T/T A/A C/C C/C C/T G/G G/G C/C A/A C/C C/C T/T T/T T/T T/T T/T G/G A/A C/C C/C A/A C/C C/C C/C C/C	A/A C/C G/G G/G C/C G/G C/C C/C C/C	A/A C/C G/G G/G G/G C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	A/A C/C G/G G/C C/T T/C G/A C/C C/C C/C C/C C/C C/C C/C C	6) 69 A/A C/C G/G G/G G/G C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C	6/6 A/A A/C G/6 G/6 C/C C/C C/C C/C G/6 C/C G/6 C/C G/6 C/C G/6 C/C G/6 C/C C/C A/A T/T T/T T/T G/6 A/A T/T T/T T/T T/T C/C C/C C/C C/C	G/G A/A C/C G/C G/C C/C C/C C/C C/C C/C C/C C/C	ТГБ G(G A/A A/A C/C G/G G/G G/C C/C C/C C/C C/C C/C C/C	ТТ G/G A/A C/C G/G G/G G/C C/C G/G C/C C/C C/C C/C C/C C/C C/C G/A G/A C/C G/A C/C G/A C/C C/C <th>, , , , , , , , , , , , , , , , , , ,</th> <th>G/G A/A A/A G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C</th>	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C
40 rs8182161 19620705 T A T/A T/T	32 33 34 35 36 37 38 39 40 41 42 43 44	15.14948/063 rs116102263 rs72761116 rs127261116 rs129826737 rs59963862 rs17135411 rs5834523 rs1992826737 rs59963862 rs17135411 rs58834533 rs190238492 rs722555 rs1273940418 rs77960132 rs13380687 rs12369751 rs12369751 rs12369751 rs12369751 rs13380456 rs11240031 rs1242039 rs12443870 rs32626272 rs12443870 rs32626272 rs12448852 rs32642072 rs12448872 rs32642072 rs12444529 rs32642072 rs12444529 rs12444529 rs12444529 rs124452072 rs12444529 rs12444529 rs12444529 rs12444529 rs1444529	1620307 1620424 1620458 1620468 1620459 1621109 1621197 1621197 162109 1621197 162109 162208 162208 162251 1622473 162251 1623473 162352 1624002 16240000000000000000000000000000	A G G G C C C T T A C C G G C T G T T T G G C C C T T T T T	T T C C T T C G T A T A C T G C C A T C G G C A G TA C C A A →	АЛТ С/С G/G G/G C/T C/C C/C C/C C/C C/C C/C C/C	А, А С/Т G/G G/G C/C C/C C/C C/C C/C C/C	Α/Α C/C G/G G/G C/C G/G C/C C/C C/C C/C C/C G/G G/G G/G G/G C/C G/G C/C C/C G/G T/T G/G T/T G/G T/T G/G C/C C/C	Α/Α C/C G/G G/G G/G C/C G/G C/C C/C C/C G/G G/G G/G G/G G/G G/G C/C C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G C/C C/C	A/A C/C G/G G/G G/G C/C C/C C/C C/C	Α/Α	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	λ/λ C/G G/G G/G G/G G/G C/C G/G C/C G/G G/A C/T G/A C/C G/A C/C G/A C/C G/A C/C C/C C/C C/C C/G/G A/G T/T T/C G/G A/G T/T C/C C/C C/C C/C		GiG G GiG G GiG G GiG G GiG G GiG G GiG G CiC C CiC C CiC C CiC GiG G CiC C CiC GiG G CiC C CiC CiC	G/G G/G A/A C/C G/C G/C G/C C/C C/C C/C C/C C/C C/C	Тіб Gi Gi Gi Gi Gi Gi Gi Gi Gi Ci Ci Ci Ci Ci Ci Ci Ci Ci Ci Ci Ci Ci	T/T G/G G/G A/A C/C G/G G/G G/G G/C C/C C/C C/C C/C C/C C/C G/A G/A C/C A/G T/T G/A G/A G/A G/G T/T G/G T/G G/A G/G T/T T/G A/G T/T T/T T/T T/T C/CA C/CA	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A C/C G/G G/G G/G C/C C/C C/C C/C C/C C/C
47 1627426 τ c 1/1 1	32 33 34 35 36 37 38 39 40 41 42 43 44 45	15.14948/063 rs116102263 rs72761116 rs127261116 rs12726117 rs19826737 rs29826737 rs29963862 rs17135411 rs58934533 rs19922642 rs7276112 rs5996382 rs17135411 rs5834533 rs199228492 rs72565 rs13230667 rs13380687 rs12380687 rs12380687 rs12380687 rs12380687 rs12380687 rs12380451 rs13380456 rs13380456 rs13280456 rs13380456 rs13380456 rs13380456 rs13380456 rs12489751 rs12489751 rs12489751 rs1248852 rs35262072 rs12488852 rs37888559 rs488852 rs59783459 rs1444579752 rs147108136	1620307 1620424 1620458 1620468 1620459 1621195 1621195 1621195 1621957 1622008 162208 162208 1622219 1622219 1622251 1623473 162352 1624102 1624102 1624423 1624423 16244243 16245244 1624524 1624524 1624524 162652 162652 162652 1626652	A G G C C C C T T A C C C C C C C C C C C C C	Т Т С С Т Т С G Т А Т А С Т G С С А Т С G G С А G ТА С С А Т А С Т G С С А Т С G G С А G ТА С С А А Т А	АЛТ С/С G/G G/G C/T G/G C/C C/C C/C C/C C/C C/C C/C	Α/Α	А/А (С (С (С (С (С (С (С (С (С (С	Α/Α	- //A -	А/А G/G G/G G/G G/C C/C C/C C/C C/T C/C C/T C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C		6.3 3 λ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 2 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3 ζ 3	GiG G GiG G GiG G GiG G GiG G GiG G GiG G GiG C CC C CC	G/G G/G A/A C/C G/C G/C G/C G/C C/C C/C C/C C/C C/C	Тіб G/G G/G G/C G/C G/C G/C G/C G/C	T/T G/G G/G A/A C/C G/G G/G G/G G/C C/C G/G G/G G/G G/G C/C G/A G/G Y/T T/T T/T T/T G/A G/G G/A G/A G/A G/A G/A G/A G/A G/A G/A	(//)/ (//)/	G/G A/A A/A C/C G/G G/G G/G G/G G/G C/C C/C C/C C/C
48 49 49 50 50 50 50 50 50 50 50 50 50	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46	15.14948/063 rs116102263 rs72761116 rs12761116 rs12761116 rs12761116 rs127621116 rs19826737 rs27481037 rs59963862 rs17135411 rs58313533 rs1990238492 rs722655 rs137940418 rs77960132 rs139300687 rs139300687 rs12969751 rs138300687 rs123969751 rs138300687 rs12369751 rs13830045127 rs138300456 rs111294003 rs124443870 rs3266072 rs12444529 rs4488852 rs59783459 rs1424457 rs117108136 rs117108136 rs117108136	1620307 1620424 1620424 1620468 1620550 162119 162119 162119 162119 1621967 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 1622092 162493 162493 162493 162493 16249784 162499 16249784 1625890 162592 1626079 1626592 1626595 1626595 1626595 1626595 1626559 1626559 1626559 16	A G G C C C C T T C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G A Т А А С	АЛТ С/С G/G G/G C/T G/G C/C C/C C/C C/C C/C C/C C/C	ΑΪΑ C/T G/G G/G G/G G/G C/C G/G C/C C/C	Α/Α Α/Α (C) (C) (G) (G) (G) (G) (C) (C) (C) (C) (C) (C) (C) (C	ΑΪΑ	- //A - //A - //C -	А/А G/G G/G G/G G/G G/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C	- X,A - C/C - G/G - G/C - C - C - C - C - C - C - C -		G/G G/G G/G G/G G/G G/G G/G G/G G/G C/C G/G G/G C/C G/G G/G C/C G/G G/G C/C G/G G/G C/C C/C G/G G/G G/G C/C C/C G/G G/G C/C C/C G/G G/G C/C C/C G/G G/G C/C C/C C/C G/G G/G C/C C/C C/C C/C C/C G/G G/G C/C	G/G G/G G/C G/C G/C G/C G/C C/C C/C C/C	ТГб G/G G/G G/G G/G G/G G/G G/G C/C C/C C/C	T/T G/G A/A C/C G/G G/G G/C C/C G/G C/C G/G G/G G/G G/A	ارا ارا G/G A/A C/C G/G G/G G/G C/C C/C C/C C/C C/C C/C C/C G/G G/G G/G C/C C/C G/G G/G C/C G/G C/C C/C C/C G/G T/T T/T G/G T/T T/G G/G T/T G/G T/T G/G T/T G/G T/T T/T C/C C/C C/C C/C T/T T/T T/T C/C C/C C/C C/C C/C C/C C/C C/C C/C	G/G G/G G/G
49 50 51 52 52 53 52 53 53 53 53 53 53 53 53 53 53	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47	15.14948/053 rs116102263 rs72761116 rs12761116 rs12761116 rs12761116 rs12761116 rs127481037 rs59826737 rs72481037 rs52481037 rs524826 rs17135411 rs58343533 rs199028402 rs22565 rs137940418 rs77960132 rs13380687 rs143380687 rs12380687 rs12380453 rs12380451 rs12380456 rs12380456 rs12380457 rs138300457 rs138300457 rs13830045127 rs13830456 rs11224023 rs12444529	1620307 1620424 1620424 1620450 1620468 1620550 1621119 1621119 16211967 1621097 1622092 1621097 1622092 1622108 1622251 1622251 16224252 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624423 1624528 1624528 1626522 1626552 162652 162652 1	А G G G C C C C C C C C C C C C C	T T C C T T C G T A T A C T G C C A T C G G C A G A T A A C C	A/T C/C G/G G/G C/T G/G C/T G/G C/T A/G C/T G/A G/A C/C G/A C/T A/G T/C G/A G/A C/T T/C G/A T/T CCCCT/C T/T T/C G/A G/A G/A G/A G/A G/A G/A T/T T/C G/A A/G T/T C/C C/A G/A G/A G/A G/A G/A G/A G/A G/A G/A G/A	А/А С/Т С/Т С/Г С/С С/С С/С С/С С/С С/С С/С	А/А	ΑΪΑ	А/А С/С G/G G/G G/G G/G C/C C/C C/C C/C	А/А G/G G/G G/G G/G C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	۸/A ۸/A ۲/A ۲/A ۲/A ۲/C ۲/T ۲/C ۲/T ۲/C ۲/T ۲/C ۲/C ۲/C ۲/C ۲/C ۲/C ۲/T ۲/T ۲/T ۲/T ۲/T ۲/T	6,6 4,A C/C 6/6 6/6 6/6 6/6 C/C 7/7 A/A C/C 6/6 C/C 7/7 A/A C/C 6/6 C/C 7/7 7/7 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C 6/6 C/C C/C	G/G G/G	G/G G/G G/C G/C G/C G/C G/C G/C C/C C/C	ТГб G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C	T/T G/G G/G A/A C/C G/G G/C C/C C/C C/C C/C G/G C/C C/C C/C G/A G/A G/A G/A G/A G/A G/A G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C G/G T/T T/G T/G G/G A/G T/T C/C G/G G/G G/G T/A G/G T/A G/G T/A G/G T/A G/G	, , , , , , , , , , , , , , , , , , ,	G/G G/G G/G G/G G/G G/G C/C C/C C/C C/C
4'9 15/39/219904 102/78/50 1A T TA/TA <	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48	15.14948/053 rs116102263 rs72761116 rs12761116 rs12761116 rs12761116 rs12761116 rs127261116 rs127261116 rs12726112 rs127135411 rs528856196 rs133411 rs5834533 rs190238423 rs123555 rs133940418 rs772655 rs133940637 rs12399051 rs12380687 rs12399051 rs12380453 rs12399075 rs12380456 rs13380451 rs060000 rs12296975 rs13380451 rs060000 rs122498751 rs13380452 rs59783459 rs12448709 rs12448709 rs12448709 rs12448709 rs12448769752 rs117108126 rs181707 rs18182161 rs117592744 rs17448844 <td< th=""><th>1620307 1620424 1620424 162050 1620468 1620550 1621119 1621119 16211967 1621097 1622092 16212967 1622092 16222092 16222092 16222092 16222092 16222092 1622408 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 162495</th><th>A G G C C C C C C T A C C C C T C A A T T C C C C</th><th>Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G A С С A Т А А С С G A</th><th>A/T C/C G/G G/G C/T G/G C/T G/G C/T G/G C/T G/G C/T G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C C/T T/C G/A G/A T/T T/C G/A G/A G/A G/A G/A G/A G/A G/G T/T T/C C/C C/A G/G T/A G/G T/A G/G G/G</th><th>ΑΪΑ</th><th>А/А (С (С (С (С (С (С (С (С (С (С</th><th>Α/Α Α/Α (C) (C) (C) (C) (C) (C) (C) (C)</th><th>А/А С/С С/С С/С С/С С/С С/С С/С</th><th>А/А С/С G/G G/G G/G C/C C/C C/C C/C C/C</th><th>A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/T C/C C/C C/C C/C C/C C/C C/C C/C C/C</th><th></th><th></th><th>G/G C</th><th>GIG GIG C/C C/C C/C G/C C/C C/C C/C C/C C/C C/</th><th>ТГб G/G G/G G/G G/C G/C G/C G/C C/C C/C C/C</th><th>T/T G/G G/G G/G G/G G/C G/G G/C C/C C/C G/G G/C C/C G/A G/G J/T G/A G/A G/G J/A G/G J/A G/G J/A G/G G/G</th><th>, , , , , , , , , , , , , , , , , , ,</th><th>G/G A/A A/A A/A C/C G/G G/G G/G G/G C/C G/G T/T T/T G/G G/G T/T G/G T/T G/G T/T G/G T/T G/G T/T A/A T/T G/G T/T A/A T/T G/G T/T A/A T/T A/A T/T A/A</th></td<>	1620307 1620424 1620424 162050 1620468 1620550 1621119 1621119 16211967 1621097 1622092 16212967 1622092 16222092 16222092 16222092 16222092 16222092 1622408 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 1624495 162495	A G G C C C C C C T A C C C C T C A A T T C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G A С С A Т А А С С G A	A/T C/C G/G G/G C/T G/G C/T G/G C/T G/G C/T G/G C/T G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C C/T T/C G/A G/A T/T T/C G/A G/A G/A G/A G/A G/A G/A G/G T/T T/C C/C C/A G/G T/A G/G T/A G/G G/G	ΑΪΑ	А/А (С (С (С (С (С (С (С (С (С (С	Α/Α Α/Α (C) (C) (C) (C) (C) (C) (C) (C)	А/А С/С С/С С/С С/С С/С С/С С/С	А/А С/С G/G G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/T C/C C/C C/C C/C C/C C/C C/C C/C C/C			G/G C	GIG GIG C/C C/C C/C G/C C/C C/C C/C C/C C/C C/	ТГб G/G G/G G/G G/C G/C G/C G/C C/C C/C C/C	T/T G/G G/G G/G G/G G/C G/G G/C C/C C/C G/G G/C C/C G/A G/G J/T G/A G/A G/G J/A G/G J/A G/G J/A G/G G/G	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A A/A C/C G/G G/G G/G G/G C/C G/G T/T T/T G/G G/G T/T G/G T/T G/G T/T G/G T/T G/G T/T A/A T/T G/G T/T A/A T/T G/G T/T A/A T/T A/A T/T A/A
50 (s)11641/2 1628225 T 6 1/T 1	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 40	15.14948/053 rs116102263 rs72761116 rs12761116 rs12448197 rs199826737 rs72761116 rs72761137 rs199826737 rs72481037 rs722565 rs13790418 rs77960132 rs13280687 rs123804563 rs123804563 rs123804563 rs123804563 rs123804563 rs123804563 rs123804563 rs123804563 rs12444870 rs52526072 rs121448270752 rs117108136 rs182164 r	1620307 1620424 1620424 1620468 162050 162119 162119 162119 162119 162109 162109 1622092 1622092 1622092 1622092 1622209 1622231 1622351 162342 162442 162442 162442 162442 162442 162442 162442 162442 162442 162442 162442 1624589 162465 1624784 1625890 1626058 1626058 1626592 16265	A G G G C C C C C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G ТА С СА А Т А А С С G А Т – Т	A/T C/C G/G G/G C/T G/G C/T G/G C/T G/G C/T G/A C/C G/A C/C G/A C/C C/C G/A C/C C/C C/C C/C G/A G/A G/A C/C C/C C/C C/C G/A G/A	Α/Α	Α/Α () () () () () () () () () ()	ΑΪΑ	А/А ССС G/G G/G G/G C/C C/C C/C C/C C/C	А/А С/С G/G G/G G/G C/C C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C T/T A/A C/C G/G C/T C/C C/C C/C C/C C/C C/C C/C C/C C/C	А/А С/С С/С С/С С/С С/С С/С С/С		G/G G/G G/G G/G G/G G/G G/G G/G	Слб Срб Срб Срб Срб Срб Срб Срб Ср	Тіб Gi Gi Gi Gi Gi Gi Gi Gi Gi Ci Ci Ci Ci Ci Ci Ci Ci Ci C	T/T G/G A/A C/C G/G G/C C/C G/A G/A C/C C/C C/A G/A C/C C/A C/C C/A C/C C/A C/C C/A C/C C/A C/C	,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,	G/G A/A A/A A/A C/C G/G G/G G/G C/C G/G C/C C/C C/C C/C C/C C/C C/C G/G C/C G/G C/C G/G C/C C/C C/T T/T T/T T/T G/G C/C C/C
51 (s36091619) 1628579 (ananangkangk	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 49	15.14948/063 rs116102263 rs72761116 rs12761116 rs12761116 rs12761116 rs127621116 rs127621116 rs127621116 rs127621116 rs127261116 rs127261116 rs127481037 rs22481037 rs22482 rs22565 rs112369751 rs112369751 rs112369751 rs112369751 rs112448370 rs2562072 rs1148769752 rs1148769752 rs1148769752 rs1124884 rs124884 rs124884 rs124884 rs124884 rs1252663	1620307 1620424 1620428 1620468 162050 162119 162119 162119 1621957 1622092 1622092 1622092 1622092 1622092 1622209 1622209 1622209 1622209 162239 1622351 162449 1624528 1624528 1624528 1624528 1624784 1625524 162552 1626058 1626058 1626058 1626552 16275555 16275555 16275555 16275555 16275555 16275555 16275555 16275555 16275555 162755555555555555555555555555555555	A G G G C C C C C C C C C C C C C C C C	T T C C T T C G T A T A C T G C C A T C G G C A G TA C A A C C G G A T T C G G C A A T A A C C G A T T T CAA	АЛТ С/С G/G G/G C/T G/G C/C C/C C/C C/C C/C C/C C/C	А/А С/Т С/Т С/Г С/С С/С С/С С/С С/С С/С С/С	ΑΪΑ	ΑΪΑ	А/А ССС G/G G/G G/G C/C C/C C/C C/C C/C	Α/Α Α/Α G/G G/G G/G G/G C/C C/C C/C C/C	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C			Gig G Gig G C/C C C C C C C C C C C C C C C C C C	Слб Срб Срб Срб Срб Срб Срб Срб Ср	ТГБ G/G A/A C/C C/C G/G G/G G/G C/C C/C C/C C/C C/C	Т/Т G/G G/G G/G G/G G/C G/G G/C G/G G/C C/C G/G G/G G/G G/G G/A C/C G/A C/C G/A C/C G/A C/C G/G T/G T/G T/G T/G T/G G/G A/G T/T C/C C/C G/G A/G T/T C/C G/G G/G G/A G/G A/G T/T T/A G/G G/A G/G G/A G/G G/A	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A C/C G/G G/G G/G G/G C/C C/C G/G C/C C/C
rs36091619 1628579 налаландалаладалаладалаладалалалаталалаладталалаладталаналадталандаланд	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 49 50	15.14948/063 rs116102263 rs72761116 rs12448197 rs19826737 rs7276116 rs1992826737 rs728261 rs1992826737 rs59963862 rs17135411 rs58343533 rs199282637 rs1725555 rs137940418 rs725651 rs137940418 rs725657 rs1320687 rs147054463 rs1090310 rs12596975 rs1330056 rs1330056 rs13130456 rs111924003 rs1243870 rs1243870 rs12443870 rs12443870 rs12443870 rs12443870 rs12443870 rs12443870 rs12438707 rs12438707 rs12438707 rs12438707 rs12438707 rs12438707 rs12438707 rs12438707 rs12438707 rs12	1620307 1620424 1620468 1620468 162050 162119 162119 162119 1621967 1621967 1621008 1621967 1622008 1622108 1622351 1622351 162363 162363 162465 162442 1624784 1624784 1624784 1624784 1624784 1624784 1625892 1626058 1626059 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626058 1626592 1626595 1627595 1627595 1627595 1627595 1627595 1627595 16	A G G G C C C C C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G TA С А А Т А А С С G А Т Т С G G С А G TA С СА А Т А А С С G А Т Т САЯ С	A/T C/C G/G G/G G/G C/T G/G C/C C/C C/C G/A G/A G/A G/A C/C G/A C/C G/A C/C C/C C/C C/C C/C G/A C/T T/C G/A C/T T/T T/T G/A G/A C/T T/T T/T G/A G/G T/T A/G G/G T/T A/G G/G T/T A/G G/G T/T A/G G/G T/T A/G	Α/Α	Α/Α Α/Α (C) (C) (C) (C) (C) (C) (C) (C)	ΑΪΑ	А/А С/С G/G G/G G/G G/G G/G G/G G/G	Α/Α () () () () () () () () () ()	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C C/C	А/А С/G G/G G/G G/C G/G G/C C/C C/T C/T C/C G/A C/C G/A C/C C/C C/C C/C C/C C/C C/C C		GiG G GiG G GiG G GiG G GiG G GiG G GiG CiC CiC CiC CiC CiC CiC CiC CiC CiC CiC	GIG GIG AIA GIG CC CC CC CC CC CC CC CC CC CC CC CC CC	ТГБ G/G A/A C/C C/C G/G G/G G/G G/G C/C C/C C/C C/C	Т/Т G/G A/A C/C G/G G/C G/C G/C C/C G/G G/C C/C G/G C/C G/A G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C G/A C/C C/C C/C C/C G/A C/C C/C C/C C/C C/C C/C C/C G/G A/G T/T T/T C/C C/C G/A G/G A/G T/T T/T T/T T/T	, , , , , , , , , , , , , , , , , , ,	G/G A/A A/A C/C G/G G/G G/G G/G C/C C/C G/G C/C C/C C/C G/G C/C G/G G/G T/T G/G C/C C/C C/C C/C G/G T/T T/T G/G C/C C/C
	32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 49 50 51	15.14948/063 rs116102263 rs72761116 rs12448197 rs72761116 rs129826737 rs59963862 rs17135411 rs589345331 rs199226737 rs59963862 rs17135411 rs58834533 rs190238422 rs722555 rs137390418 rs77960132 rs13380687 rs13380687 rs12369751 rs13380661 rs12369751 rs13380456 rs112360751 rs13380456 rs11294003 rs1242003 rs124003 rs12443870 rs12443870 rs12488852 rs9783459 rs1448769752 rs117108136 rs8182161 rs11252744 rs11252744 rs11252744 rs11252744 rs11252744 rs1124884 rs11252744 rs1252744 <td< th=""><th>1620307 1620424 1620424 1620424 162050 162119 162119 1621495 1621967 1621967 1621967 1622008 1622321 1622495 1622325 1623251 162443 162442 162442 162442 162442 162444 1624528 1624724 1624724 1624724 162529 1626058 1626058 1626058 1626592 1627598 1627</th><th>A G G G C C C C C C C C C C C C C C C C</th><th>Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G ТА С С А А Т А А С С G А Т Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т Т О О О О О О О О О О О О О О</th><th>А/Т С/С G/G G/G C/T G/G C/T G/G C/C C/C C/C C/C G/A G/A G/A G/A C/C C/G/A G/A G/A G/G T/T T/C C/C C/G G/G T/T G/G T/T T/A G/G T/T T/A G/G T/T T/T T/A</th><th>Α/Α Α/Α C/T C/T G/G G/G G/G C/C G/G C/C C/C C/C C/C G/G G/G G/G C/C G/G C/C G/G C/C G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G T/T T/T T/T T/T T/T T/T</th><th>ΑΪΑ</th><th>ΑΪΑ</th><th>А/А С/С G/G G/G G/G G/G G/G G/G C/C C/C</th><th>А/А () () () () () () () () () ()</th><th>A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G G/G</th><th></th><th></th><th>G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C</th><th>С, ССС, ССС, ССС, ССС, ССС, ССС, ССС,</th><th>Тіб Gi Gi Gi Gi Gi Gi Gi Gi Gi Gi</th><th>T/T G/G G/G G/G G/G G/G G/G G/G G/G C/C G/G G/G C/C C/C G/A G/A G/A G/A G/A C/C G/A G/A C/C C/C C/C G/A G/A C/C G/A G/G A/G T/T T/T T/T T/T T/G G/A G/A G/A</th><th>, , , , , , , , , , , , , , , , , , ,</th><th>G/G A/A C/C G/G G/G G/G G/G G/G C/C C/C G/G G/G C/C C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C</th></td<>	1620307 1620424 1620424 1620424 162050 162119 162119 1621495 1621967 1621967 1621967 1622008 1622321 1622495 1622325 1623251 162443 162442 162442 162442 162442 162444 1624528 1624724 1624724 1624724 162529 1626058 1626058 1626058 1626592 1627598 1627	A G G G C C C C C C C C C C C C C C C C	Т Т С С Т С Т Т С G Т А Т А С Т G С С А Т С G G С А G ТА С С А А Т А А С С G А Т Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С G А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т С О О А Т Т Т О О О О О О О О О О О О О О	А/Т С/С G/G G/G C/T G/G C/T G/G C/C C/C C/C C/C G/A G/A G/A G/A C/C C/G/A G/A G/A G/G T/T T/C C/C C/G G/G T/T G/G T/T T/A G/G T/T T/A G/G T/T T/T T/A	Α/Α Α/Α C/T C/T G/G G/G G/G C/C G/G C/C C/C C/C C/C G/G G/G G/G C/C G/G C/C G/G C/C G/G C/C C/C C/C C/C C/C C/C G/G C/C G/G T/T T/T T/T T/T T/T T/T	ΑΪΑ	ΑΪΑ	А/А С/С G/G G/G G/G G/G G/G G/G C/C C/C	А/А () () () () () () () () () ()	A/A C/C G/G G/G C/C C/C C/C C/C C/C C/C G/G G/G			G/G G/G G/G G/G G/G G/G G/G G/G C/C C/C	С, ССС, ССС, ССС, ССС, ССС, ССС, ССС,	Тіб Gi Gi Gi Gi Gi Gi Gi Gi Gi Gi	T/T G/G G/G G/G G/G G/G G/G G/G G/G C/C G/G G/G C/C C/C G/A G/A G/A G/A G/A C/C G/A G/A C/C C/C C/C G/A G/A C/C G/A G/G A/G T/T T/T T/T T/T T/G G/A G/A G/A	, , , , , , , , , , , , , , , , , , ,	G/G A/A C/C G/G G/G G/G G/G G/G C/C C/C G/G G/G C/C C/C C/C G/G C/C G/G C/C G/G C/C G/G C/C C/C

rs368082114	1628604	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C
 rs138003160	1629061	T	С	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T
 rs573551047 rs11867080	1629964	T	c	T/C	T/T	T/T	T/T	T/T	T/T	T/T	CA/CA T/T	T/T	T/T	T/T	T/T	CA/CA T/T	CA/CA T/T	T/T
 rs3743507	1630381	G	А	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G
 rs56218869	1631294	A	G	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/G	A/A	A/A	A/A	A/A	A/A
 rs114623241 rs140950029	1631536	G	ACT	G/A A/ACT	G/G A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/A	G/G A/ACT	G/G A/A	G/G A/A	G/G A/ACT	G/G A/A	G/G A/ACT	G/G A/A	G/G A/A
rs187641585	1631762	G	А	A/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
 rs117406049	1631809	Т	G	G/G	T/T	T/T	T/T	T/T	T/T	T/T	T/G	T/T	T/T	T/T	T/T	T/G	T/T	T/T
 rs72761124	1632448	c	T	C/C	C/C	C/C	C/C	C/C C/C	C/C C/C	C/C C/C	C/C C/C	C/C C/C	C/C	C/C	C/C	C/C	C/C	C/C C/C
rs9934342	1632965	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
 rs9927202	1633100	A	G	A/G	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A
rs147086637 rs8055710	1633446	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	A/A G/G	G/G	G/A	G/G	G/A	G/G	G/G
rs28406883	1633946	G	с	G/C	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
 rs34762152	1634385	C	T	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
 rs55695276	1635137	A	G	A/G	A/A	A/A	A/A	G/G A/A	A/A	G/A A/A	G/G A/A	G/A A/A	A/A	A/A	A/A	A/A	G/A A/A	G/G A/A
rs28647246	1635752	т	с	T/C	T/T	T/T	T/T	T/T	T/T	T/T	т/т	T/T	T/T	T/T	т/т	T/T	T/T	T/T
 rs28608068	1635872	C	Т	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs2236268	1636549	c	Â	C/A	C/C	c/c	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs71860883	1636628	GCA	G	GCA/G	GCA/GCA	GCA/GCA	GCA/GCA	GCA/G	GCA/G	GCA/G	GCA/G	GCA/GCA	GCA/GCA	G/G	GCA/GCA	GCA/GCA	GCA/G	GCA/GCA
 rs76842429	1636896	C	T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
rs77750249	1636935	Ť	G	T/T	т/т	т/т	т/т	т/т	т/т	т/т	T/G	T/T	т/т	T/G	т/т	T/G	T/T	T/T
rs574261663	1637667	А	G	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A
 rs35972580	1637962	C	T G	C/C A/A	C/C A/A	C/C A/A	C/C A/A	C/C	C/C	C/C	C/C	C/T	C/C A/A	C/C	C/C A/A	C/C	C/C	C/C A/A
 rs180758804	1638450	c	т	c/c	c/c	c/c	c/c	c/c	C/T	C/C	C/T	c/c	c/c	c/c	c/c	c/c	c/c	c/c
rs145693119	1638528	А	TAATAAT	A/A	A/A	A/A	A/A	AATAATAAT	A/A	AATAATAA	A/A	A/A	A/A	A/A	A/A	A/A	AATAATAAT	A/A
 rs145693119 rs72761127	1638528	A	AATAATA/ T	A/A C/T	A/A C/C	A/A C/C	A/A C/C	A/A	A/A	A/A	TAATAATA	A/A C/C	A/A C/C	A/A C/C	A/A C/C	A/A	A/A	A/A C/C
 rs9924141	1638989	т	c	T/C	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T
 rs142607578	1639029	G	A	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G
 rs12447357 rs112431294	1639793 1639860	C	G T	C/G A/A	C/C A/A	C/C A/A	C/C A/A	C/C A/T	C/C A/A	C/C A/A	C/G A/A	C/C A/A	C/C A/A	C/G A/A	C/C A/A	C/G A/A	C/C A/A	C/C A/A
rs72761128	1640035	т	с	T/C	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	т/т	T/T	т/т	т/т	T/T
 rs758621945	1640201	С	CA	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/CA	C/C	C/C
 rs11641248	1640301	c	G T	C/G C/C	C/C	C/C	c/c	C/C	C/C C/C	c/c	C/C	C/C C/C	c/c c/c	С/С	C/C	C/C C/T	c/c	c/c
rs536399679	1641983	G	А	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G
rs116859784	1642026	G	A	G/A	G/G	G/A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
rs201188361 rs2273679	1642423	c	T	С/С	C/C	C/C	c/c	C/I C/C	C/C C/C	c/c	C/C	c/c c/c	C/C C/C	С/С	c/c	С/С	c/c	C/C C/C
 rs112565100	1642660	т	с	T/C	T/T	T/T	T/T	T/T	т/т	т/т	T/C	т/т	T/T	T/C	т/т	T/C	т/т	т/т
 rs9927226	1642679	G	A	G/A	G/G T/T	G/G T/T	G/G	G/G	G/G T/T	G/G T/T	G/G	G/G T/T	G/G T/T	G/G	G/G	G/G	G/G T/T	G/G T/T
rs796687743	1643316	ст	c	C/T	C/T	C/T	C/T	CT/C	CT/CT	CT/CT	CT/CT	ст/ст	C/T	C/T	C/T	CT/CT	CT/C	CT/CT
 rs187540964	1643455	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
 rs12446250	1643552	C	T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
rs116910642	1644032	ĉ	т	c/c	c/c	c/c	c/c	c/c	c/c	c/c	C/C	c/c	C/C	C/C	c/c	C/C	c/c	C/T
rs536466763	1644227	G	GT	G/G	G/G	G/G	G/G	G/GT	G/G	G/GT	G/GT	G/G	G/G	G/G	G/G	G/G	G/GT	G/GT
 rs5/833692 rs2755183	1644227	G	GT	G/G C/G	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/GT C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C	G/G C/C
rs2941956	1644764	A	G	A/G	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A
 rs9933780	1644808	C	G	C/G	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
 rs112191146 rs2755184	1645264	T	A C	G/A T/C	G/G T/T	G/G T/T	G/G T/T	G/G T/C	G/G T/T	G/G T/C	G/G T/C	G/G C/C	G/G T/T	G/G C/C	G/G T/C	G/G C/C	G/G T/C	G/G T/T
rs548658471	1645604	G	GA	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/GA	G/GA	G/G	G/G	G/G	G/GA	G/G	G/GA
 rs17135416	1645745	Т	C	T/C	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T
rs35576736	1646018	C	T	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	T/T	C/C	C/T	C/T	C/T	C/T	C/C
rs2667669	1646543	G	А	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
rs193299834	1646787	C A	T	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs143684159	1646973	c	A	c/c	C/A	C/C	C/C	C/C	C/C	c/c	c/c	c/c	C/C	c/c	C/C	c/c	c/c	c/c
rs6600156	1648126	А	С	A/C	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/A	A/C	A/C	A/A	A/A	A/A
 rs6600156 rs2974845	1648126 1648162	A T	C C	A/A T/C	A/A T/T	A/A T/T	A/A T/T	A/C T/C	A/A T/T	A/C T/C	A/A T/C	C/C	A/A T/T	A/A C/C	A/A T/C	A/C	A/C T/C	A/A T/T
rs4786368	1648638	G	c	G/C	G/G	G/G	G/G	G/C	G/G	G/C	G/C	c/c	G/C	c/c	G/C	c/c	G/C	G/G
rs78305748	1648685	G	С	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/C	G/G	G/G	G/C	G/G	G/C	G/G	G/G
 rs186995221 rs201340184	1649287	A T	G	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/T	A/A T/C	A/G T/T	A/A T/C	A/A T/T	A/A T/T	A/A T/T
rs180744609	1649476	G	т	G/G	G/T	G/T	G/T	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
rs769942411	1650461	AT	A	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT	AT/A	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT	AT/AT
 rs137922934	1650463	TTA	T	TTA/TTA	TTA/TTA	TTA/TTA	TTA/TTA	TTA/TTA	TTA/TTA	TTA/TTA	TTA/TA	TTA/TTA	TTA/TTA	TTA/TA	ΠΑ/ΠΑ ΠΑ/ΠΑ	TTA/TA	TTA/TTA	TTA/TTA
 rs34665250	1650464	TA	т	TA/T	TA/TA	TA/TA	TA/TA	TA/T	TA/T	TA/T	TA/T	TA/T	TA/TA	TA/TA	TA/TA	TA/TA	TA/T	TA/T
rs34665250	1650464	AAAAAA T	AAAAAA/	AAAA/TAAA T/T	AAAA/TAAA T/T	AAAA/TAAA T/T	AAAA/TAAA T/T	AAAA/TAAA	AAAA/TAAA T/T	AAAA/TAAA			AAAA/TAAA T/T		AAAA/TAAA T/T		AAAA/TAAA	
 rs760111992	1650894	CA	C	CA/CA	CA/CA	CA/CA	CA/CA	CA/C	CA/C	CA/C	CA/C	CA/C	CA/CA	CA/CA	CA/CA	CA/C	CA/C	CA/C
 rs2667684	1650946	А	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/G	G/G	A/G	G/G	A/G	A/A
 rs35379437	1651258	c	Т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	c/c	C/C
 rs146128830	1652418	c	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	c/c	C/C	C/T	C/C	C/C
 rs142257947	1652546	G	А	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G
 rs2236269	1652614	T A	A	T/A	T/T A/A	T/T A/A	T/T A/A	T/A	T/T	T/A	T/A	A/A	T/T A/A	A/A A/G	T/A A/A	A/A	T/A	T/T
rs140148755	1652680	c	A	c/c	C/C	C/C	C/C	C/A	C/C	C/C	c/c	C/C	C/C	C/C	C/C	C/C	c/c	C/C
 rs2667681	1653206	С	Т	C/T	C/C	C/C	C/C	C/T	C/C	C/T	C/T	T/T	C/C	T/T	C/T	T/T	C/T	C/C
 rs75640119 rs537190496	1653223 1653487	C C	G	C/C	C/C	C/C	C/C C/C	C/C C/C	C/C C/C	C/C	C/G C/C	c/c c/c	C/C	C/G	C/C C/C	C/G C/C	c/c	C/C C/C
 rs1570210	1654265	G	A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/A	G/G	G/G	G/A	G/G	G/A	G/G	G/G
 rs552736270	1654275	C	GTGT,CGT	C/C	C/C	C/C	C/C	C/C	C/CGT	C/C	C/CGTGT	C/CGT	C/C	C/C	C/C	C/CGTGT	C/CGTGTGT	C/CGT
rs/52/6217	1654319	G	TGAGG	G/G	G/G T/T	G/G T/T	G/G T/T	G/G T/TGAGG	G/G T/T	G/G T/TGAGG	G/G T/TGAGG	G/G GAGG/TGAG	G/T	GAGG/TGAG	T/TGAGG	G/G GAGG/TGAG	G/G	G/G T/T
rs10701133	1054330		TOAGO	1/10/10/														

								-										
rs138669173	1654525	С	G	C/C	C/C	C/C	C/C	C/C	C/C	C/G	C/C	C/C	C/C	C/C	C/C	C/C	C/G	C/C
rs7197288	1654753	С	A	C/A	C/C	C/C	C/C	C/A	C/C	C/A	C/A	A/A	C/C	A/A	C/A	A/A	C/A	C/C
rs2667679	1654946	G	A	G/A	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G
rs56359342	1654948	G	A	G/A	G/G	G/G	G/G	G/A	G/G	G/A	G/G	A/A	G/G	G/A	G/A	G/A	G/A	G/G
rs117745556	1655073	т	С	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/C	T/T	T/T	T/C	T/T	T/C	T/T	T/T
rs35220693	1655439	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
rs6600157	1656321	G	A	G/A	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/G
rs142169797	1656564	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs2859310	1656765	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A
rs542474562	1656927	GCACA	G	CACA/GCA	CCACA/GCAC	CACA/GCAC	CACA/GCAC	CACA/GCA	CACA/GCAC	CACA/GCAC	CACA/GCAC	CACA/GCAG	CACA/GCAC	CACA/GCA	CCACA/GCAC	CACA/GCAC	CACA/GCAC	GCACA/G
rs7192480	1657012	С	т	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	T/T	C/C	C/T	C/T	C/T	C/T	C/C
rs34935994	1657034	CAT	С	CAT/C	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT	CAT/CAT
rs1894649	1657284	С	G	C/G	C/C	C/C	C/C	C/G	C/C	C/G	C/G	G/G	C/C	G/G	C/G	G/G	C/G	C/C
rs743963	1657350	A	т	A/T	A/A	A/A	A/A	A/T	A/A	A/T	A/A	T/T	A/A	A/T	A/T	A/T	A/T	A/A
rs3784840	1657754	С	т	C/C	C/C	C/C	C/C	C/T	C/C	C/T	C/C	T/T	C/C	C/T	C/T	C/T	C/T	C/C
rs743964	1657853	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
rs763152	1658056	т	С	T/C	T/T	T/T	T/T	T/C	T/T	T/C	T/C	C/C	T/T	C/C	T/C	C/C	T/C	T/T
rs763153	1658086	G	С	G/G	G/G	G/G	G/G	G/G	G/G	G/G	G/C	G/G	G/G	G/C	G/G	G/C	G/G	G/G
rs35888283	1658101	TAG	т	TAG/T	TAG/TAG	TAG/TAG	TAG/TAG	TAG/T	TAG/TAG	TAG/T	TAG/TAG	T/T	TAG/TAG	TAG/T	TAG/T	TAG/T	TAG/T	TAG/TAG
rs9937922	1658154	С	G	C/G	C/C	C/C	C/C	C/G	C/C	C/G	C/G	G/G	C/C	G/G	C/G	G/G	C/G	C/C
rs115729443	1658454	С	т	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/T	C/C	C/C	C/T	C/C	C/T	C/C	C/C
 rs11438070	1658632	С	CA	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs28519681	1659128	т	С	T/C	T/T	T/T	T/T	T/C	T/T	T/C	T/C	C/C	T/T	C/C	T/C	C/C	T/C	T/T
rs28633318	1659723	т	G	T/G	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T	T/T
rs1057983	1660274	A	G	A/G	A/A	A/A	A/A	A/G	A/A	A/G	A/G	G/G	A/A	G/G	A/G	G/G	A/G	A/A
rs150781872	1661491	С	CA	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C	C/C
rs2859314	1661701	A	G	A/G	A/A	A/A	A/A	A/A	A/A	A/G	A/G	A/G	A/A	G/G	A/A	A/G	A/G	A/A
rs553272330	1661814	CA	С	CA/CA	CA/CA	CA/CA	CA/CA	CA/CA	CA/C	CA/C	CA/C	CA/C	CA/CA	CA/CA	CA/CA	CA/C	CA/CA	CA/C

Table S6. Haplotype analysis for ~100kb around *IFT140* on chromosome 16.

SNP analysis at the *IFT140* locus for carrier individuals of the tandem duplication and in controls sequenced either by WGS or Target Exome Sequencing (TES, by the Leber panel which include the full *IFT140* gene). In total, 6 carrier individuals from 3 families were analysed (family A, B and C) and indicated. Affected patients are depicted with an "*". Six control samples are also shown. For each individual, SNP detected in *IFT140* were extracted and listed. Homozygous SNP are shown in green (reference allele) or in blue (alternative allele) and heterozygous SNP in orange. The duplication region is highlighted in grey.

e pezez



Figure S5 AluJb and AluJr alignment

AluJb and AluJr sequences were extracted from IGV (Thorvaldsdottir, et al., 2013) at the breakpoints of the tandem duplications. Pairwise alignment of the nucleotide sequences and percent identity has been computed using Jalview (Waterhouse, et al., 2009).

John Wiley & Sons, Inc.

Technique	Individuals	CANOES	Pindel	CNVnator
Ciliome panel	D-II.1	DUP, intron26-intron30	67 DEL, 13 INV, 35 SI	NA
Ciliome panel	K-II.2	2 DEL, 3DUP (DUP, intron27-exon31)	36 DEL, 1 INV, 6 SI, 1 TD	NA
Leber panel	B-I.1	DUP, intron26-intron30	DUP, intron26-intron30 (675 DEL, 70 INV, 233 SI, 5 TD)	NA
Leber panel	B-I.2	0	(688 DEL, 62 INV, 211 SI, 2 TD)	NA
Leber panel	B-II.1	DUP, intron26-intron30	DUP, intron26-intron30 (595 DEL, 55 INV, 193 SI, 2 TD)	NA
Leber panel	C-II.1	DUP, intron26-intron30	DUP, intron26-intron30 (842 DEL, 120 INV, 417 SI, 5 TD)	NA
Leber panel	I-II.1	1 DEL	(659 DEL, 115 INV, 339 SI, 2 TD)	NA
WES	A-II.1	0	10 DEL, 3 INV, 10 SI, 1 TD	NA
WES	A-II.2	DUP, intron27-intron30	13 DEL, 3 INV, 9 SI	NA
WGS	A-II.1	DUP, exon26-intron31	14 DEL, 3 INV, 4 SI	0
WGS	A-II.2	DUP, exon26-intron31	10 DEL, 2 INV, 3 SI	0
WGS	A-I.2	DUP, exon26-intron31	17 DEL, 1 INV, 2 SI	0
WGS	A-I.1	0	53 DEL, 1 INV, 1 SI	0

 Table S7. Summary of the CNV detected in *IFT140* by using 3 different tools in 13 individuals.

CNV detection has been performed on 13 individuals sequenced with 4 different approaches: the Ciliome panel, the Leber panel, the WES and the WGS. Three tools have been run with their default options: CANOES (v1.0), Pindel (v0.2.5b9) and CNVnator (v0.3.3) (Abyzov, et al., 2011; Backenroth, et al., 2014; Ye, et al., 2009). The number of CNV detected in *IFT140* are reported here, as well as the presence or not of the tandem duplication of exons 26 to 30. It is to notice that Pindel simultaneously calls small indel and structural variations, increasing thus the number of detected variants. NA, not applicable; DUP, duplication; DEL, deletion; INV, inversion; SI, short insertion; TD, tandem duplication.

SUPPLEMENTARY REFERENCES

Abyzov A, Urban AE, Snyder M, Gerstein M. 2011. CNVnator: an approach to discover, genotype, and characterize typical and atypical CNVs from family and population genome sequencing. Genome Res 21(6):974-84.

Adzhubei IA, Schmidt S, Peshkin L, Ramensky VE, Gerasimova A, Bork P, Kondrashov AS, Sunyaev SR. 2010. A method and server for predicting damaging missense mutations. Nature Methods 7(4):248-9.

Backenroth D, Homsy J, Murillo LR, Glessner J, Lin E, Brueckner M, Lifton R, Goldmuntz E, Chung WK, Shen Y. 2014. CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Res 42(12):e97.

Bayat A, Kerr B, Douzgou S, Study DDD. 2017. The evolving craniofacial phenotype of a patient with Sensenbrenner syndrome caused by IFT140 compound heterozygous mutations. Clin Dysmorphol 26(4):247-251.

Beheshtian M, Saee Rad S, Babanejad M, Mohseni M, Hashemi H, Eshghabadi A, Hajizadeh F, Akbari MR, Kahrizi K, Riazi Esfahani M and others. 2015. Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. Arch Iran Med 18(11):776-85.

Bifari IN, Elkhamary SM, Bolz HJ, Khan AO. 2016. The ophthalmic phenotype of IFT140related ciliopathy ranges from isolated to syndromic congenital retinal dystrophy. Br J Ophthalmol 100(6):829-33.

DePristo MA, Banks E, Poplin R, Garimella KV, Maguire JR, Hartl C, Philippakis AA, del Angel G, Rivas MA, Hanna M and others. 2011. A framework for variation discovery and genotyping using next-generation DNA sequencing data. Nat Genet 43(5):491-8.

Gerber S, Alzayady KJ, Burglen L, Bremond-Gignac D, Marchesin V, Roche O, Rio M, Funalot B, Calmon R, Durr A and others. 2016. Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. Am J Hum Genet 98(5):971-980.

Grampa V, Delous M, Zaidan M, Odye G, Thomas S, Elkhartoufi N, Filhol E, Niel O, Silbermann F, Lebreton C and others. 2016. Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. PLoS Genet 12(3):e1005894.

Helm BM, Willer JR, Sadeghpour A, Golzio C, Crouch E, Vergano SS, Katsanis N, Davis EE. 2017. Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic mutation in IFT140 that causes Mainzer-Saldino syndrome. Hum Genomics 11(1):16.

Huerta-Cepas J, Szklarczyk D, Forslund K, Cook H, Heller D, Walter MC, Rattei T, Mende DR, Sunagawa S, Kuhn M and others. 2016. eggNOG 4.5: a hierarchical orthology framework with improved functional annotations for eukaryotic, prokaryotic and viral sequences. Nucleic Acids Res 44(D1):D286-93.

Hull S, Owen N, Islam F, Tracey-White D, Plagnol V, Holder GE, Michaelides M, Carss K, Raymond FL, Rozet JM and others. 2016. Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140. Invest Ophthalmol Vis Sci 57(3):1053-62.

Khan AO, Bolz HJ, Bergmann C. 2014. Early-onset severe retinal dystrophy as the initial presentation of IFT140-related skeletal ciliopathy. J AAPOS 18(2):203-5.

Kumar P, Henikoff S, Ng PC. 2009. Predicting the effects of coding non-synonymous variants on protein function using the SIFT algorithm. Nature protocols 4(7):1073-81.

Li H, Durbin R. 2010. Fast and accurate long-read alignment with Burrows-Wheeler transform. Bioinformatics 26(5):589-95.

Pena-Padilla C, Marshall CR, Walker S, Scherer SW, Tavares-Macias G, Razo-Jimenez G, Bobadilla-Morales L, Acosta-Fernandez E, Corona-Rivera A, Mendoza-Londono R and others. 2016. Compound heterozygous mutations in the IFT140 gene cause Opitz trigonocephaly C syndrome in a patient with typical features of a ciliopathy. Clin Genet.

Perrault I, Saunier S, Hanein S, Filhol E, Bizet AA, Collins F, Salih MA, Gerber S, Delphin N, Bigot K and others. 2012. Mainzer-Saldino syndrome is a ciliopathy caused by IFT140 mutations. Am J Hum Genet 90(5):864-70.

Schmidts M, Frank V, Eisenberger T, Al Turki S, Bizet AA, Antony D, Rix S, Decker C, Bachmann N, Bald M and others. 2013. Combined NGS approaches identify mutations in the intraflagellar transport gene IFT140 in skeletal ciliopathies with early progressive kidney Disease. Hum Mutat 34(5):714-24.

Schwarz JM, Rodelsperger C, Schuelke M, Seelow D. 2010. MutationTaster evaluates disease-causing potential of sequence alterations. Nat Methods 7(8):575-6.

Thorvaldsdottir H, Robinson JT, Mesirov JP. 2013. Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. Brief Bioinform 14(2):178-92. Waterhouse AM, Procter JB, Martin DM, Clamp M, Barton GJ. 2009. Jalview Version 2--a multiple sequence alignment editor and analysis workbench. Bioinformatics 25(9):1189-91.

Weisschuh N, Mayer AK, Strom TM, Kohl S, Glockle N, Schubach M, Andreasson S, Bernd A, Birch DG, Hamel CP and others. 2016. Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. PLoS One 11(1):e0145951.

Xu M, Yang L, Wang F, Li H, Wang X, Wang W, Ge Z, Wang K, Zhao L, Li H and others. 2015. Mutations in human IFT140 cause non-syndromic retinal degeneration. Hum Genet 134(10):1069-78.

Ye K, Schulz MH, Long Q, Apweiler R, Ning Z. 2009. Pindel: a pattern growth approach to detect break points of large deletions and medium sized insertions from paired-end short reads. Bioinformatics 25(21):2865-71.

'e perez