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Translocation Breakpoint at 7q31 Associated with Tics:

Further Evidence for *IMMP2L* as a Candidate Gene for Tourette Syndrome

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20 **ABSTRACT**

21 Gilles de la Tourette syndrome is a complex neuropsychiatric disorder with a strong
22 genetic basis. We identified a male patient with Tourette syndrome-like tics and an
23 apparently balanced *de novo* translocation [46,XY,t(2;7)(p24.2;q31)]. Further analysis
24 using array comparative genomic hybridisation (CGH) has revealed a cryptic deletion at
25 7q31.1 to 7q31.2. Breakpoints disrupting this region have been reported in one isolated
26 and one familial case of Tourette syndrome. In our case, *IMMP2L*, a gene coding for a
27 human homologue of the yeast inner mitochondrial membrane peptidase subunit 2, was
28 disrupted by the breakpoint on 7q31.1, with deletion of exons 1-3 of the gene. The
29 *IMMP2L* gene has previously been proposed as a candidate gene for Tourette syndrome,
30 and our case provides further evidence of its possible role in the pathogenesis. The
31 deleted region (7q31.1. to 7q31.2) of 7.2Mb of genomic DNA also encompasses
32 numerous genes including *FOXP2*, associated with verbal dyspraxia, and the *CFTR*
33 gene.

34 **Keywords: Tourette syndrome; tics; *IMMP2L*; 7q31.1 region; array-CGH;**
35 ***FOXP2*; breakpoint mapping**

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41 **INTRODUCTION**

42 Gilles de la Tourette syndrome (GTS [MIM 137580]) is a neuropsychiatric disorder
43 manifested by multiple, involuntary motor and vocal movements (tics), that fluctuate in
44 severity. The motor tics are usually the initial symptom of the condition with onset
45 occurring usually between the ages of 2 and 14 years. Echolalia, grunting and coprolalia
46 can develop as the disease progresses. GTS was previously considered to be a rare
47 disorder, but population-based surveys estimate the prevalence at around 1/1000 males
48 and 1/10000 females.¹ GTS has been reported with other behavioural disorders such as
49 obsessive compulsive disorder (OCD), attention deficit hyperactivity disorder (ADHD),
50 and conduct disorder-oppositional defiant disorder.²

51 It is thought that GTS is inherited in an autosomal dominant manner with
52 variable expression and reduced penetrance, as around 10% of patients have a family
53 history of the same condition.³ Several genome-wide linkage studies have failed to
54 identify any candidate genes, but the largest genetic linkage study so far in over 2000
55 individuals showed strong evidence of linkage at 2p23.2, 3p, 3q, 5p, 6p, and 14q.⁴
56 Boghosian-Sell et al. (1996) described a family affected by Tourette syndrome
57 segregating with a balanced chromosomal translocation [t(7;18)]. The Tourette
58 syndrome in this family showed variable expression and reduced penetrance.⁵ No genes
59 in the region of the breakpoint on chromosome 18 have been implicated in the aetiology
60 of Tourette syndrome. The breakpoint on chromosome 7 was further mapped to 7q22-
61 q31, between two genetic markers, D7S515 and D7S522. Kroisel et al. (2001) reported
62 a 13 year old male with Tourette syndrome, minor physical anomalies, and a de novo
63 partial duplication of chromosome 7q [dup(7)(q22.1-q31.1)].⁶ Molecular analysis of this

64 case by Petek et al. (2001) revealed that the duplication was inverted. The distal
65 breakpoint occurred between the same two genetic markers that defined the region
66 disrupted in the case by Boghosian-Sell et al. (1996), raising the possibility of a
67 candidate gene for GTS at 7q31. Further studies identified that a novel gene, *IMMP2L*
68 (MIM 605977), was interrupted by both the breakpoint in the duplicated fragment and
69 the insertion site in 7q31.⁷

70 We describe detailed molecular genetic analysis of an 18 year old male with a *de*
71 *novo* translocation t(2;7)(p24.2;q31) who developed motor tics at the age of 13 years
72 and has significant speech and language impairment. The breakpoints are further
73 characterised and the *IMMP2L* gene is found to be disrupted and partially deleted by the
74 translocation, along with a cryptic 7.2Mb deletion involving a number of genes.

75 **SUBJECT and METHODS**

76 ***Case Report***

77 The 18 year old male proband is the third child of healthy nonconsanguineous White
78 British parents. He was born at term with a birth weight of 2980 g (9-25th centiles). He
79 sat unsupported at age 9 months and walked at age 17 months: later than his normal
80 siblings. At age 3 years 6 months he had poor speech development with only two word
81 sentences. His oromotor control was poor resulting in constant severe dribbling. He
82 attended a school for children with learning difficulties. At age 13 years he developed a
83 movement disorder consisting of head and eye turning to the right, facial grimacing and
84 puffing of his cheeks. He was partially able to control this, but it appears to be
85 worsening with age. He was seen by a paediatric neurologist who made a clinical

86 diagnosis of tics rather than dystonia. He continues to have moderate learning
87 difficulties and severe speech difficulties, with verbal dyspraxia and his speech is barely
88 comprehensible to strangers. There is no family history of mental retardation, speech
89 delay or movement disorders.

90 On examination at age 18 years his weight is between the 25-50th centiles and
91 height between the 50-75th centiles. He has no facial dysmorphism, but has an
92 intermittent divergent squint and hypoplastic 5th toenails. Neurological findings include
93 the tics, normal tone and power in all four limbs but brisk deep tendon reflexes, and a
94 broad based, unsteady gait. An electroencephalogram showed a non specific slight
95 increase in background activity, but no focal or generalised seizure activity. A CT scan
96 of the brain showed an enlarged cisterna magna, but was otherwise normal.

97 *Cytogenetic Analysis*

98 Routine cytogenetic analyses were performed on metaphase chromosomes prepared
99 from peripheral blood leukocytes using standard methods.

100 *Array Studies*

101 Genomic DNA was extracted from peripheral blood using the AutoPure extraction
102 system (Qiagen) following the manufacturers instructions. DNA concentration and
103 purity ($A_{260-280} \sim 1.8-2.0$) was determined using a NanoDrop spectrophotometer (Labtech
104 International). Samples were labelled with Cy3 and Cy5 using a NimbleGen Dual
105 Colour DNA Labelling kit according to manufacturers instructions. 500ng genomic
106 DNAs were hybridised to NimbleGen 12x135K arrays (Roche NimbleGen) and arrays
107 washed post-hybridisation according to manufacturer's instructions. Arrays were

108 scanned using a GenePix 4000B scanner and analysed in Nexus Copy Number v3.0
109 (BioDiscovery, Inc.), using the Rank Segmentation algorithm at default settings.
110 Oligonucleotide probes were mapped to NCBI36.

111 *Fluorescent In Situ Hybridisation (FISH)*

112 DNAs for bacterial artificial chromosome (BAC) FISH probes were obtained from
113 Roswell Park Cancer Institute, USA and labelled in house with Cy3 (GE Healthcare)
114 using the Bioprime labelling kit (Invitrogen). Hybridisation and washing was carried
115 out using standard protocols. Images were visualised using an Olympus BX50
116 microscope and captured and processed using the Isis software package (Metasystems).

117 *MPLA*

118 Multiplex ligation-dependent probe amplification (MLPA) analysis of the CFTR gene
119 was carried out using the MRC-Holland P091 kit according to the manufacturer's
120 instructions and using a Tetrad PCR machine (MJ Research). Capillary electrophoresis
121 was carried out on an ABI 3130 and analysed using the Soft Genetics Gene Marker
122 v1.70 program.

123 **RESULTS**

124 *Identification of an apparently balanced translocation*

125 Cytogenetic analysis identified a reciprocal translocation between the short arm of
126 chromosome 2 at band p24.2 and the long arm of chromosome 7 at band q31 (Fig. 1).
127 This was determined to be a *de novo* finding following analysis of parental samples.
128 The chromosome 7 breakpoint occurred in the same band as a previously described

129 duplication that disrupted the *IMMP2L* gene and implicated this gene as a candidate for
130 a role in the pathogenesis of GTS.⁷

131 ***Identification of deletion at chromosome 7 breakpoint using array-CGH***

132 Array analysis showed a 7.25Mb deletion from 7q31.1 to 7q31.2 at the breakpoint of the
133 t(2;7) translocation. The deleted region contained 338 oligonucleotide probes genomic
134 co-ordinates 7:110702484 to 117947839) (Fig. 2a & 2b). The proximal breakpoint of
135 the deletion occurs within introns 2-3 of the *IMMP2L* gene, and exons 1 to 3 are
136 deleted. Exons 4 to 6 remain intact. The deleted region also contains 21 HGNC mapped
137 genes (Table 1) including the OMIM Morbid entries *FOXP2*, *MET* and *CFTR*. Parental
138 samples were not analysed using array-CGH as the translocation was determined to be a
139 *de novo* finding following analysis of parental chromosomes.

140 ***FISH confirmation of partial deletion of IMMP2L***

141 FISH analysis using RP11-452K21 (co-ordinates 7:110396144 to 110575013)
142 overlapping the 3' end of *IMMP2L* showed a signal present on both the derivative and
143 normal chromosomes 7 confirming disruption of *IMMP2L* and deletion of the 5' end of
144 the gene only (Fig. 2c). RP11-148C1 (co-ordinates 7:110833766 to 111014525,
145 overlapping the 5' end of *IMMP2L*) showed loss of signal on the derivative
146 chromosome 7 (no movement of the signal to the derivative chromosome 2 was
147 observed). Clear signal for this BAC was observed on the normal chromosome 7
148 confirming deletion of the 5' end of one copy of *IMMP2L* (Fig. 2d). The FISH results
149 therefore confirm the proximal breakpoint assigned by microarray analysis to lie
150 between 110.58 and 110.83Mb, within introns 2-3 of *IMMP2L*.

151 ***Comparison of copy number imbalance overlapping *IMMP2L* using Ensembl and***
152 ***DECIPHER resources***

153 Comparison of the deleted region in this case to other copy number imbalances
154 involving the *IMMP2L* gene was carried out using the Database of Chromosomal
155 Imbalance and Phenotype in Humans using Ensembl Resources (DECIPHER)(Patient
156 ID in DECIPHER: 248350). This identified nine additional cases with imbalance in this
157 region only (DECIPHER entries 800, 801, 804, 813, 919, 1296, 1299, 1303 and 2391).
158 Only one of these had motor and vocal tics as a clinical symptom (DECIPHER entry
159 919) which is the case reported by Petek et al. (2001) of a duplication of 7q22.1 to
160 7q31.1 with the distal breakpoint between exons 3 and 4 of *IMMP2L*.⁷ Of note there are
161 several entries of smaller copy number variation (CNV) within the Database of
162 Genomic Variants (DGV) within the *IMMP2L* gene (Fig. 2b). Most of these encompass
163 introns 2-3 but several also appear to include exons 1-3.

164 ***Confirmation of deletion of the *CFTR* gene by MLPA***

165 Analysis of the *CFTR* gene using MLPA showed a heterozygous deletion of all exons
166 of the gene at 7q31.2 confirming the array and FISH results. The most centromeric of
167 the probes shown to be deleted was located within the *ASZI* gene that sits 58kb
168 upstream of the *CFTR* gene and the most telomeric probe deleted is located within the
169 *CTTNBP2* gene that sits 58kb downstream of the *CFTR* gene (data not shown).

170 **DISCUSSION**

171 Chromosome abnormalities that segregate with a disease phenotype can facilitate the
172 identification of disease loci and genes. Patients with an abnormal clinical phenotype

173 and an apparently balanced *de novo* chromosome rearrangement on routine karyotyping,
174 have been shown to have submicroscopic imbalances on further analysis. Using CGH
175 arrays, De Gregori et al. (2007) found 40% of patients with an abnormal phenotype and
176 an apparently balanced translocation, were in fact unbalanced.⁸ The majority of patients
177 with Tourette syndrome have a normal karyotype and of the various chromosomal
178 abnormalities reported in association with Tourette syndrome, only the translocation
179 cases reported by Boghosian-Sell et al. (1996) and Kroisel et al. (2001), show
180 cosegregation with features associated with Tourette syndrome.^{5,6} Together, they
181 provided evidence that a gene located in the breakpoint region at 7q31 could be
182 involved in the development of Tourette syndrome. Previously reported cases with 7q
183 duplications have not shown any signs of Tourette syndrome, which again supports the
184 notion of involvement of gene(s) at the breakpoints rather than genes within the
185 duplicated region (with haploinsufficiency as the mechanism).

186 Petek et al. (2001) identified a novel gene, *IMMP2L*, disrupted by the
187 breakpoints in their case of an inverted duplication of 7q31, and proposed this gene to
188 be a candidate for GTS.⁷ The *IMMP2L* (inner mitochondrial membrane peptidase-2-
189 like) gene spans 860kb of genomic DNA, consisting of six exons encoding a 175 amino
190 acid protein. The gene shows evolutionary conservation. The human protein is 41%
191 identical to the yeast protein and 90% identical to the mouse protein. It is thought to
192 function as a catalytic subunit of the mitochondrial inner membrane peptidase (IMP)
193 complex. The IMP complex generates mature, active proteins in the mitochondrial
194 intermembrane space by proteolytically removing the mitochondrial targeting
195 presequence of nuclear-encoded proteins.⁹ Although little is known about the exact

196 function of the protein, of note, other mitochondrial proteins (e.g. COXII) have been
197 shown to be involved in neurodegenerative and neuropsychiatric disorders.¹⁰⁻¹²

198 Family based association studies by Díaz-Anzaldúa et al. (2004) in a French-
199 Canadian GTS syndrome sample showed significant association with the region
200 previously described by Petek et al. (2001) in their case with the inverted duplication of
201 7q31. They found a significant association close to the first duplication breakpoint with
202 markers D7S523 and D7S522 (situated 1.2 and 5 Mb distal to this breakpoint), and
203 marker D7S1516 (situated within the *IMMP2L* gene).¹³ A further study in 2007 by
204 Petek et al. screening 39 unrelated patients with GTS, for mutations in *IMMP2L*,
205 detected no coding-region variants, and so failed to provide further evidence that
206 *IMMP2L* mutations are a common cause of GTS. However, the gene may be involved at
207 a more subtle level or in just a small proportion of cases. They concluded that to
208 exclude this gene from a pathogenic role in GTS, studies screening for mutations in the
209 intronic, promoter, enhancer and other regulatory elements is required.¹⁴

210 A number of cases of deletions and duplications overlapping the 7q31.1 region
211 (and including *IMMP2L*) have been reported in the DECIPHER database but only one
212 case has documented vocal and motor tics (DECIPHER entry 919) as part of the
213 phenotype. There could be a number of explanations for this. Firstly, some of these
214 cases are younger than 3 years old and may be too young to have yet developed features
215 of the condition. Both our case and that reported by Kroisel et al. (2001) developed
216 Tourette syndrome after age 11 years. Secondly, the absence of GTS could also be
217 explained by the variable penetrance of the condition. Lastly, the cases in DECIPHER
218 do not involve disruption of *IMMP2L*, but all involve complete deletion leading to

219 haploinsufficiency for *IMMP2L*. Our case (with deletion of exons 1-3 out of 6) and the
220 translocation case studied by Petek et al. (2001) provide evidence of disruption of
221 *IMMP2L*. A truncated gene and the inability to transcribe a full length mRNA, along
222 with the production of a defective protein may be responsible for the appearance of
223 features of GTS, and our case provides further evidence for this. This may be an
224 important pathogenetic mechanism rather than deletion or duplication of the entire gene.
225 Reported cases of CNV within *IMMP2L* also exist in the DGV, although, given variable
226 penetrance associated with GTS and the lack of phenotypic data available for “healthy
227 control cohorts” generally assessed for studies of “normal” variation recorded in the
228 DGV, it is not possible to exclude involvement of *IMMP2L* in GTS on the basis of these
229 cases. These points also highlight the importance of follow up of all cases with
230 overlapping deletions and duplications encompassing *IMMP2L* gene, not only to allow
231 early detection and management of features of GTS, but also allow further
232 understanding of the genetic mechanisms by which the *IMMP2L* gene is implicated in
233 the pathogenesis of GTS.

234 Our case has significant verbal dyspraxia and speech difficulties. He sometimes
235 omits the end sound in words and has difficulty producing the sound of some
236 consonants. He has problems producing words with three or more syllables especially
237 when used in sentences and conversation. Array CGH analysis in our case identified the
238 *FOXP2* gene (MIM 605317) to be completely deleted. The *FOXP2* gene encodes a
239 transcription factor thought to regulate gene expression in defined areas of the
240 developing neural, lung, cardiovascular and intestinal tissue, and was the first gene to be
241 implicated in a developmental communication disorder. Abnormalities of *FOXP2* are
242 associated with difficulties in the learning and production of sequences of oral

243 movement, which impair speech, known as developmental verbal dyspraxia. Affected
244 individuals have variable levels of impairment in expressive and receptive language,
245 sometimes extending to problems with production and comprehension of grammar.
246 Hurst et al. (1990) described a large multigenerational family with a severe speech and
247 language disorder inherited in an autosomal dominant manner.¹⁵ A similar speech and
248 language disorder was presented by Lai et al. (2000) in a patient with a *de novo*
249 balanced reciprocal translocation t(5;7)(q22;q31.2).¹⁶ They mapped the 7q31.2
250 breakpoint to a single clone, containing several exons of the *FOXP2* gene, showing
251 disruption of this gene in their case. They also identified a point mutation in the *FOXP2*
252 gene in the family reported by Hurst et al. (1990), which co-segregated perfectly with
253 the speech and language disorder in this family.¹⁷ Case reports of deletions within the
254 7q31 region including *FOXP2*,¹⁸ and studies showing mutations within *FOXP2* in
255 isolated cases of verbal dyspraxia,¹⁹ have further supported the evidence of this gene
256 being important for normal speech and language development. It is very likely that the
257 speech impairment, verbal dyspraxia, and possibly oropharyngeal dyspraxia and
258 dribbling, seen in our case are due to haploinsufficiency of this gene.

259 The *IMMP2L* gene (along with numerous other neurodevelopmental genes) has
260 also recently been implicated in structural variation studies carried out on cohorts of
261 patients with autistic spectrum disorder,²⁰ and attention-deficit hyperactivity disorder.²¹
262 Our case does not display any of the classical features of autism, except speech
263 impairment (which is likely to be accounted for by deletion of the *FOXP2* gene), or
264 attention-deficit hyperactivity disorder. Autism or autistic spectrum disorder (ASD
265 [MIM 209850]) is a severe neurodevelopmental disorder that presents at an early age
266 and persists throughout life. Individuals display impairments in social interaction and

267 communication coupled with repetitive and/or obsessive-compulsive behaviours. Data
268 from family and twin studies suggests a strong genetic component, with the likelihood
269 of numerous genes contributing to the pathogenesis in an additive manner. Genetic and
270 cytogenetic studies have provided evidence to suggest one or more autism susceptibility
271 genes within the 7q22-q31 region.²² Hutcheson et al. (2003) further defined this region
272 to a 3 cM region on 7q31-q33 between markers D7S496 and D7S523, containing
273 several interesting functional candidate genes including some neuronally expressed
274 genes: *NRCAM* (MIM 601581) and *LRRN3*.²³ Neither of these genes is within our
275 deleted region, but *LRRN3* is located centromeric to the proximal deletion breakpoint
276 and very near the last few exons of the *IMMP2L* gene.

277 Our case does have moderate mental retardation and this could be accounted for
278 by haploinsufficiency of an as yet unknown mental retardation gene within the deleted
279 region. Some of the genes within the deleted region may be good candidate genes for a
280 role in the pathogenesis of mental retardation (e.g. *DOCK4*, *GPR85*, *MET*, *WNT2*, and
281 *CTTNBP2*). The *DOCK4* gene (MIM 607679) has been shown to regulate dendritic
282 growth in neuronal cell lines in rats.²⁴ A recent case-control study in families linked to
283 the autism susceptibility locus 1 (*AUTS1*) detected single nucleotide polymorphisms
284 (SNP) and copy number variations (CNV) within *DOCK4* and *IMMP2L* implicating
285 them in autism susceptibility.²⁰ A *DOCK4* deletion segregating in a family with
286 dyslexia, and undetectable in >2500 controls, suggested *DOCK4* may have a role in the
287 aetiology of dyslexia.²⁵ The *GPR85* gene (MIM 605188) is predominantly expressed in
288 the central nervous system, especially structures exhibiting high levels of plasticity (e.g.
289 hippocampal dentate gyrus), it has high evolutionary conservation across three
290 mammalian species,²⁶ and has been implicated as a potential risk factor for psychiatric

291 disorders.²⁷ The *MET* gene (MIM 164860) encodes for a receptor tyrosine kinase and
292 participates in neocortical and cerebellar growth and maturation, as well as immune and
293 gastrointestinal functions. In 204 families with autism, Campbell et al. (2006) found a
294 significant association between autism and a G-C transversion in the promoter of the
295 *MET* gene. Functional assays showed the C allele resulted in a 2 fold decrease in *MET*
296 promoter activity and altered binding of specific transcription factor complexes.²⁸ The
297 WNT family of genes influences the development of the central nervous system and a
298 mouse knockout of the *DVLI* gene (a member of a gene family essential for the function
299 of the WNT pathway) exhibited a behavioural phenotype characterised by diminished
300 social interaction. Wassink et al. (2001) screened the *WNT2* gene (MIM 147870) in a
301 large number of autistic probands and found two families containing a nonconservative
302 coding sequence variant that segregated with autism in those families.²⁹ Cheung et al.
303 (2001) showed high levels of expression of *CTTNBP2* (MIM 609772) in the brain and
304 screened it for mutations in 90 unrelated individuals with autism, and identified two
305 variants.³⁰ It is possible though that speech and language impairment and autistic traits
306 may be influenced by a single gene or multiple interacting genes within this region.

307 The *CFTR* gene (MIM 602421) is also located within the deleted region in our
308 patient, therefore making him a carrier of Cystic fibrosis (CF) (CF [MIM 219700]). This
309 will have implications for future offspring and the necessity to offer carrier testing to
310 any future partners. This case, in addition to providing supportive evidence for
311 disruption of *IMMP2L* gene function causing GTS, has highlighted some very important
312 genetic counselling issues for the patient: 1) standard “balanced” translocation (and the
313 risk of larger chromosomal imbalance in any offspring), 2) offspring risk for GTS, 3)

314 offspring risk for mental retardation, 4) offspring risk for verbal dyspraxia, and 5)
315 implications of being a CF carrier.

316 **CONCLUSION**

317 Our case adds to evidence from previous reports of the importance of genes within the
318 7q31 region, particularly the association of *IMMP2L* with GTS, and *FOXP2* with verbal
319 dyspraxia. There is evidence for considerable genetic heterogeneity in GTS, and the
320 number of cases attributable to abnormalities of *IMMP2L* is yet to be determined.
321 Further studies are required to determine the role of *IMMP2L* in the aetiology of GTS.
322 We also highlight the value of array CGH to obtain a cytogenetic diagnosis in an
323 apparently balanced translocation with an abnormal phenotype, and the identification of
324 new candidate gene.

325 **ACKNOWLEDGEMENTS**

326 We are particularly grateful for the parents of the case who supported our publication of
327 their son's case to benefit other families.

328 **CONFLICT OF INTEREST**

329 The authors declare that there are no conflicts of interest.

330 **WEB RESOURCES**

331 Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl

332 Resources (DECIPHER), <http://decipher.sanger.ac.uk>

333 Database of Genomic Variants (DGV), <http://projects.tcag.ca/variation/>

334 Ensemble, <http://www.ensembl.org/index.html>

335 Online Mendelian Inheritance in Man (OMIM), <http://www.ncbi.nlm.nih.gov/OMIM/>.

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426

427 **FIGURE LEGENDS**

428 **Figure 1:** Partial G-banded karyogram showing t(2;7)(p24.2;q31) translocation

429 (*breakpoints arrowed*).

430

431 **Figure 2:**

432 **a)** Chromosome 7 log 2 ratio displayed in Nexus Copy Number v 3.0 showing 7.2Mb

433 deletion (*indicated with red bar*).

434 **b)** Zoomed proximal 7q31.1 region showing breakpoint at 110,702,484 Mbp (*red dotted*

435 *line*) within IMMP2L. Exons 1-2 and exons 1-3 are deleted from the IMMP2L-201 and

436 IMMP2L-202 transcripts respectively.

437 **c)** Confirmatory FISH analysis showing intact copies of RP11-452K21 (110.39-

438 110.58Mb).

439 **d)** Confirmatory FISH analysis showing deletion of one copy of RP11-148C1 (110.83-

440 111.01Mb).

441 Red signals represent target BAC clones (*locations arrowed*) and green signals are

442 control probe (CEP7, Abbott Vysis).

Table 1: HGNC mapped genes in deletion interval 110702484 -117947839 (NBC1 36)

Gene	Start (Mbp)	End (Mbp)	Description and aliases
<i>IMMP2L</i>	110090346	110948739	IMP2 inner mitochondrial membrane peptidase-like (<i>S. cerevisiae</i>). Aliases: IMP2
<i>DOCK4</i>	111153404	111431423	dedicator of cytokinesis 4. Aliases: FLJ34238, KIAA0716
<i>IFRD1</i>	111877751	111903480	interferon-related developmental regulator 1. Aliases: PC4, TIS7
<i>C7orf53</i>	111908282	111918171	chromosome 7 open reading frame 53. Aliases: FLJ39575
<i>TMEM168</i>	112193032	112217714	transmembrane protein 168. Aliases: DKFZp564C012,FLJ13576
<i>GPR85</i>	112507704	112515015	G protein-coupled receptor 85. Aliases: SREB2
<i>PPP1R3A</i>	113301622	113346300	protein phosphatase 1, regulatory (inhibitor) subunit 3A. Aliases:
<i>FOXP2</i>	113842288	114121063	forkhead box P2. Aliases: CAGH44
<i>MDFIC</i>	114349445	114446501	MyoD family inhibitor domain containing. Aliases: HIC
<i>TFEC</i>	115362448	115458103	transcription factor EC. Aliases: TCFEC, TFECL
<i>TES</i>	115637817	115686071	testis derived transcript (3 LIM domains). Aliases: DKFZP586B2022, TESS-2, TESTIN
<i>CAV2</i>	115926532	115935830	caveolin 2. Aliases: CAV
<i>CAV1</i>	115952075	115988466	caveolin 1, caveolae protein, 22kDa. Aliases:
<i>MET</i>	116099682	116225676	met proto-oncogene (hepatocyte growth factor receptor). Aliases: HGFR, RCCP2
<i>CAPZA2</i>	116289799	116346548	capping protein (actin filament) muscle Z-line, alpha 2. Aliases: CAPZ, CAPP2
<i>ST7</i>	116380617	116657393	suppression of tumorigenicity 7. Aliases: TSG7, SEN4, ETS7q, HELG, RAY1, FAM4A
<i>WNT2</i>	116704518	116750579	wingless-type MMTV integration site family member 2. Aliases: IRP
<i>ASZ1</i>	116790512	116854779	ankyrin repeat, SAM and basic leucine zipper domain containing 1. Aliases: Orf3, GASZ, ALP1
<i>CFTR</i>	116907253	117095955	cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7).
<i>CTTNBP2</i>	117137944	117300797	cortactin binding protein 2. Aliases: KIAA1758, Orf4
<i>LSM8</i>	117611322	117620114	LSM8 homolog, U6 small nuclear RNA associated (<i>S. cerevisiae</i>). Aliases: YJR022W
<i>ANKRD7</i>	117651953	117669977	ankyrin repeat domain 7. Aliases: TSA806



