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## Title page

### Care management in a French cohort with Down syndrome from the AnDDI-Rares/CNSA study.

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## **Abstract (250 words)**

Down syndrome (DS) is a genetic neurodevelopmental disorder. In individuals with DS, a multidisciplinary approach to care is required to prevent multiple medical complications. The aim of this study was to describe the rehabilitation, medical care, and educational and social support provided to school-aged French DS patients with varying neuropsychological profiles. A mixed study was conducted.

Quantitative data were obtained from a French multicentre study that included patients aged 4 to 20 years with diverse genetic syndromes. Qualitative data were collected by semi-structured face-to-face interviews and focus groups. Ninety-five DS subjects with a mean age of 10.9 years were included. Sixty-six per cent had a moderate intellectual disability (ID) and 18.9 % had a severe ID. Medical supervision was generally multidisciplinary but access to medical specialists was often difficult. In terms of education, 94% of children under the age of six were in typical classes. After the age of 15, 75% were in medico-social institutions. Analysis of multidisciplinary rehabilitation conducted in the public and private sectors revealed failure to access physiotherapy, psychomotor therapy and occupational therapy, but not speech therapy. The main barrier encountered by patients was the difficulty accessing appropriate facilities due to a lack of space and long waiting lists. In conclusion, children and adolescents with DS generally received appropriate care. Though the management of children with DS has been improved considerably, access to health facilities remains inadequate.

**Key words:** Down syndrome; intellectual deficiency; multidisciplinary care; social support; educational support

## **Main Text**

### **Introduction**

Down syndrome (DS) is a chromosomal abnormality caused by the presence of a third copy of chromosome 21, and it is one of the most common congenital genetic syndromes. In France, the live birth prevalence of DS is 1 per 2000 [1]. Despite the effect of prenatal screening policies, the number of children born with DS has remained stable due to the increase in average maternal age at childbirth; additionally, the overall prevalence of individuals with DS has increased in recent years along with life expectancy [1-4].

DS is characterized by intellectual disability (ID), which is generally mild to moderate, hypotonia, hyperlaxity and delayed psychomotor development [5]. These patients may have other comorbidities such as congenital heart disease (40-50% of cases), gastrointestinal malformations (especially duodenal atresia), visual or auditory deficits (congenital cataract, strabismus, perceptive deafness), sleep apnoea syndrome, thyroid disorders, immune deficiency, epilepsy, behavioural disorders, or motor impairment [6-7]. The profiles of children with DS can be very different depending on the level of impairment and the associated comorbidities.

The variability of DS makes the management of the disorder very complex, which implies that it should be multidisciplinary and based on a holistic approach. There is a need to focus on rehabilitation (which includes speech therapy, psychomotor therapy, physiotherapy, psychological counselling and occupational therapy), medical care, schooling and social support, and ideally all of these aspects should be coordinated. In France, 131 reference centres and 501 competence centres for rare diseases received accreditation for the French Ministry of Health for this type of coordination between 2004 and 2007. Additional aid can also be provided by specific patient support groups whose purpose is to help individuals with DS and their families obtain support and recognition (i.e. Trisomie 21 France).

In addition to creating a French reference centre for rare diseases, the French law n°2005-102, dated 11<sup>th</sup> February 2005, provides guidelines for the care of disabled persons and the assistance necessary to manage the consequences of a disability of any origin or nature, whatever the age or the lifestyle of the individual [8]. This law has also led to the creation of a public administrative centre for disability in each French department (*Maison Départementale des Personnes Handicapées - MDPH*), which makes decisions regarding financial compensation, equipment needs, and special needs in schooling.

Rehabilitation (speech therapy, psychomotor therapy, physiotherapy, psychological counselling, and occupational therapy) can be offered at the child's home, at school, in a private practice or through organisations offering rehabilitation services, educational support and home care (*Service d'Education Spéciale et de Soins A Domicile - SESSAD*). Otherwise, it takes place in a medico-social institution (centre for developmentally disabled children). Children with ID can also attend typical classes in mainstream schools, often with the support of an educational assistant, or in special needs classes with fewer children, specialised teachers and adapted teaching materials. They can also be enrolled in medico-social institutions, which combine recreational activities, social support, rehabilitation and medical monitoring (the services can be very different from one institution to another) [9]. The current situation can be confusing for individuals with disabilities and their families due to the unequal distribution of resources across the French territory [10]

To our knowledge, there are currently no published studies depicting the detailed management of patients with DS in France. Previously published data regarding patients with disabilities provided a general overview, without specifying the type of care or support according to the degree of ID or the disease [11-12]. In this context, we aimed to assess the medical care, rehabilitation, and social and educational support that is available depending on the degree of ID in children with DS using a qualitative and quantitative study.

## Methods

We conducted a mixed study, which is well-suited for studying complex multidisciplinary health pathways [13]. The specific design for our research was an explanatory sequential design (QUAN → qual) in which qualitative data are collected after quantitative data in order to provide a better understanding and add more depth to the explanation of the quantitative data [14].

### *Quantitative study: data collection and analysis*

The population of individuals with DS in this study was extracted from a national database which includes several other rare diseases such as DiGeorge syndrome, Williams syndrome, Angelman syndrome, Smith-Magenis syndrome, and fragile X syndrome. This national cross-sectional study financed by the National Solidarity Fund for Autonomy (*Caisse Nationale de Solidarité pour l'Autonomie - CNSA*), called the CNSA Study, was designed to describe the medical care, and social and educational support provided to school-aged persons with diverse chromosomal abnormalities, and to investigate a possible correlation with the degree of intellectual disability. Nine reference centres for rare diseases, which are part of the AnDDI-Rares network for developmental diseases in France, were enrolled in the study. The inclusion period was from mid-2011 to the end of 2012. Inclusion criteria were the presence of a proven chromosomal abnormality by standard karyotyping or array-CGH, an age between 4 and 20 years, national health insurance system coverage, and signed informed consent from the parents to take part in the study. Exclusion criteria included involuntary hospitalization or guardianship. Parents were invited to take part in the study either by the reference centres, who contacted individuals by mail and phone, or through patient support groups. The project was approved by the Ethics Committee of Robert Debré hospital in Paris, France, where the data were centralized and processed.

Once the child was included in the study, the parents received a standardized questionnaire to fill in. The questionnaire collected data regarding neonatal features (sex, weight, height, etc.), demographics (age, family structure, number of siblings, etc.), medical follow-up (physicians involved in monitoring, treatment, etc.), rehabilitation (type, frequency, etc.), education (type of schooling), social context (types of financial compensation, parental occupation, etc.) and satisfaction. The parents and affected children were then asked to spend one day at their reference centre. During the visit, the questionnaire was checked with the help of an in-house investigator. Then, an overall assessment of the children's intellectual performance comprising different sub-domains (verbal, non-verbal, working memory and processing speed) was conducted with Wechsler's scales (according to age: WPPSI-III, WISC-IV or WAIS-III) by a neuropsychologist recruited for the study. Regardless of the type of scale, ID was defined by a total intelligence quotient (TIQ) score under 70, with profound ID corresponding to a TIQ score under 19, severe ID between 20-34, moderate ID between 35-49, and mild ID between 50-69.

### *Statistical analyses*

Qualitative variables are presented as numbers (percentages), and quantitative variables as means and standard deviations (SD) when normally distributed, or medians and ranges otherwise. Qualitative variables were compared using Fisher's exact test and, quantitative variables were compared using the Mann-Whitney U-test. Correlations between quantitative variables were assessed using Spearman rank correlation coefficients. A  $p$ -value below 0.05 was considered significant. All analyses were done using SAS version 9.4.

### *Qualitative study: data collection and analysis*

In addition to the quantitative approach, a qualitative study called T21Qual was conducted in Burgundy between January and February 2017. Semi-structured face-to-face



interviews and focus groups were conducted with health and medico-social professionals in order to collect their views on the diversity of the existing structures in France and on the current problems related to the management of children and teenagers with DS. A maximum variation sampling approach was used for recruitment (variety of jobs and workplaces).

An interview guide was developed from a literature review and in collaboration with a health economist, a social worker and a paediatrician. Participants were invited to discuss five themes: their work, coordination with other professionals, healthcare pathways, financial compensation and their overall experience.

Participants were chosen according to their occupation and their workplace (private sector, SESSAD, medico-social institution, MDPH, Regional Health Agency (ARS)) and were contacted via email. Interviews lasting approximately 60 minutes were conducted and recorded (audio only) at the participants' places of work. Participants provided oral consent, and they were informed that they could end their participation at any time.

In total, six interviews (3 face to face interviews and 3 focus groups) were conducted during the months of January and February 2017 with 13 professionals from the region of Burgundy (Table 1). The professionals are referred to in the results by the number showed in Table 1 (e.g. P1 for participant 1). The interviews were discontinued when data saturation was achieved. The interviews were anonymised and transcribed by one researcher [15]. The data were then analysed thematically: Relevant quotes were identified in the interview transcripts and attached to a code. A code is a tag used to retrieve and categorize similar data. Then, codes were put into context with each other to develop the main themes of the data set. In the results part, the most illustrative quotes will be presented.

## **Results**

### *Population description*

The population characteristics are presented in Table 2. All of the individuals in our study had DS secondary to an additional independent chromosome 21 due to an accidental

non-disjunction during meiosis. Ninety-five patients with DS aged between 4 and 20 were included. They represented 13.6 % of the CNSA study population. The mean age was 10.9 years (SD=4.8) with a male predominance (56.8%). Most of the children had moderate (66.3%) or severe intellectual disability (18.9%). The parents came from every type of socio-professional category (data not shown). Psychomotor development is described in Table 3. There was a global delay in all the stages of psychomotor development, which was heterogeneous. The definitive genetic diagnosis was made before one month of age for all but two patients. For twelve patients (12.6%), the diagnosis was made during antenatal screening. In the other cases, DS was suspected due to specific facial features (63.6% of the cases), neonatal hypotonia (30.3% of the cases) or congenital heart disease (7.6% of the cases).

#### *Medical care*

Medical monitoring was usually multidisciplinary, involving 5.3 specialists on average (SD=2.0) and up to 10 specialists. Most of the children were followed by an ophthalmologist (87.0%), a general practitioner (83.7%), a dentist (70.7%) and an ear-nose-throat specialist (69.6%) (Table 4). The average number of medical consultations per year was 11.8 (SD=7.5), regardless of the degree of ID ( $p=0.76$ ) or age ( $p=0.82$ ) (results not shown).

The qualitative results suggested that centres of excellence such as University hospitals are needed for the medical follow up (P1: *“the challenge is to have centres of excellence which could care for children with developmental disorders”* ; P4 : *“it is precisely children who will have a follow up in University hospital”*). Also, it was considered difficult to gain access to specialists but not more than for the general population (P1: *“it [the difficulties] would be access to specialists, but that's basically the case for everyone and especially for the paediatric population and the disabled paediatric population”*). Sometimes, the follow up included an institution, private practice and a university hospital (P4 : *“they have consultations in private practice in addition to the institutional care, and then every 6 months or every year,*

*there is a specialist doctor at the hospital who sees them again”).*

## *Rehabilitation*

The average number of rehabilitation sessions per child was 2.4 (SD=1.4) per week. It was significantly different depending on the age group ( $p=0.022$ ), with 2.8 (SD=1.5) sessions for children aged between 4 and 6 and 1.6 (SD=0.8) session per week for those aged between 15 and 20. However, there was no significant difference depending on the degree of ID ( $p=0.58$ ) (data not shown). The participants in the qualitative study considered that the children had either not enough rehabilitation sessions or that the sessions were not long enough (P4: *“there are no answers about the intensity of the rehabilitation [...], for me it’s not intense enough”*). Nevertheless, they stressed the importance of early intervention for the overall development of the children (P6: *“from the moment we intervene at an early stage, I would say... the support is much better, bears fruit over time”* P11 : *“because in the two children who did not benefit from early intervention, the difference is noticeable”*).

Every child had at least one type of rehabilitation, which was also observed by the health professionals from the qualitative study (P4: *“they all have rehabilitation”*; P2: *“the fact that there is a child who stays at home without...without any care, without any follow-up, without...I don’t think that exists anymore”*; P3: *“I think that all children with DS have rehabilitation”*).

The type of rehabilitation depended on age, as shown in Figure 1. The most frequent types of rehabilitation were speech therapy, psychomotor therapy, physiotherapy and psychological counselling in 93.3%, 47.2%, 22.5% and 19.1% of the children, respectively. Only 5.6% received occupational therapy. The proportion of patients who received speech therapy was stable in all age categories. However, the percentage decreased with age for psychomotor therapy (38.5% patients aged between 4 and 6 and 10.3% of patients aged over 15) and physiotherapy (only 5.3% of patients aged over 15). Rehabilitation sessions

took place in the public sector (37.0%), in the private sector (37.0%) or both (26.0%) (data not shown). These results were confirmed by the qualitative study. For instance, P1 explained: *“he could have speech therapy here or psychomotor therapy there, [...] it’s the person who chooses his own pathway... I need this, or I need that, etc.”* and concerning the sector, P8 added *“it’s true that with the tremendously long waiting lists, people are turning to private sector”*.

### *Education*

In the qualitative study, the participants discussed the effect of the French law from 2005 and particularly its effect on education. More specifically, this law addressed the inclusion of children with disabilities in typical schools (P2: *“before the 2005 law, schooling was less of a priority”*). However, one participant added this was not always the case: P4: *“since the law of 2005, young people with Down’s syndrome and any other disability should be normally in a local school, and often it is not done”*. They highlighted the decisive role of the patient support groups in this process (P5 *“they pushed a lot and this inclusion law came about because the patient support groups were very militant and made things change”*)

In the quantitative study, only one child did not go to school. The type of schooling by age group is described in Figure 2. Ninety-four percent of the children between 4 and 6 years attended typical classes. Between 6 and 8 years, most of the children who were still in typical classes were in kindergarten. After 8 and until 15 years of age, children were mostly attending either special needs classes or were enrolled in a medico-social institution. After 15 years, 75% were in a medico-social institution.

The type of schooling was significantly different according to age ( $p=0.001$ ) and the degree of ID ( $p=0.0213$ ). All patients with a profound ID and 68.4% of patients with severe ID were in special schools. Children with moderate ID were educated almost equally either in typical classes (39.1%), special needs classes (26.6%) and medico-social institutions

(34.4%). An educational assistant was present for 64.3% of the children on average 5.1 (SD=2.1) half-days a week (data not shown).

On this subject, professionals highlighted the importance of tailoring each child's path through the educational system. However, two pivotal points were commonly seen: kindergarten until 6 or 7 years, a special needs classes until the age of 10 or 11, and then on to a medico-social institution (P3: *"kindergarten, then in a special needs class in primary and secondary education, and during this pathway, in primary or secondary education, sooner or later there is the medico-social institution, depending on the degree of intellectual disability"* or P4 : *"In kindergarten, 100% of young people with DS are in typical classes [...], "in primary education, [...] 90% of young people with DS go to special needs classes [...] "then it is special needs classes in secondary education [...] but almost 50% of young people with DS are directed to medico-social institutions and they are not given the opportunity to go to the special needs classes"*).

### *Social*

Sixty-nine percent of mothers stated that they changed from a full-time job to a part-time job because of their child's disability, compared to 3.5% for fathers. Twenty-four percent of parents reported that the grandparents provided regular caregiving. Twenty-two percent of parent also reported having to deal with global constraints related to the child's disability, and 65% of them had moved as a result.

The qualitative study confirmed these results. P5 explained that *"an isolated parent with a disabled child has difficulties finding babysitting or transporting him to get medical care. So, this means reorganizing your working hours. It affects everything"*.

### *Recognition and financial compensation*

The average time period between the diagnosis of DS and official recognition of the

disability by a public administrative centre for disability was 8.3 months (SD = 12.9). The total time was more than one year for 19.2% of individuals. The average time period between official recognition and receiving financial support was 10.6 months (SD = 33.3). Furthermore, 4.5% of individuals received no financial compensation (data not shown).

Administrative procedures were described by the participants as often complicated and time consuming, even if the diagnosis was made at birth as explained by P6: *“it is not an easy procedure to get the disability recognized. [...] The child was born with a proven disability..., with the time to go through the required formalities; it could last, maybe 6 months, or a year”*

The qualitative study also indicated that families had a poor understanding of their rights. This could explain the absence of financial compensation in some families (P10: *“there are parents who don’t know that they can receive financial compensation”*).

For the participants, these difficulties could lead to parental exhaustion: P5: *“the parents are exhausted”*; P5: *“they have to do all the procedures, make all the trips”*; P1: *“I see many parents struggling”*; P1: *“Parents are in survivor mode”*.

### *Access to health facilities*

Access to health facilities was not evaluated in the quantitative study. However, it was one of the themes that was discussed during the interviews and focus groups. All participants reported long waiting times and difficulties gaining access to adapted rehabilitation and education due to the lack of capacity in the public sector (P7: *“the waiting time for the SESSAD is related to the fact that children followed by the SESSAD are waiting several years to get access to a medico-social institution. Also, the teenagers in medico-social institutions are waiting to go into an adult institution. So, that’s it, we have this specialized pathway and the whole thing is blocked”*; P1: *“medico-social institution do not have enough capacity, that’s the reality”*; P2: *“Now we’re reaching a saturation of [the health facilities]”*

*capacity”; P3: “we would like to have more facilities”; P4: “here, there are only two facilities and they are full; P5: “you have to wait until there is a spot, P10: “obviously, there is a lack of capacity”; P12: we know very well that there is long waiting list, people wait for 2 or 3 years”).*

## **Discussion**

The originality of this study lies in its mixed approach. We present here the first analyses from a national study that aims to describe the medical care, rehabilitation, financial, social and educational support provided with regard to the degree of intellectual disability (ID) in patients with DS. The qualitative study makes it possible to better understand the health pathways of these patients, and it also provides context for the quantitative results. In this way, the interviews confronted the perspectives of professionals in the care sector with the results of the national study.

In our population, only 12.6 % of patients were diagnosed with DS through antenatal screening. This can be explained by the high rate of abortion after antenatal diagnosis [1-2]. In France in 2016, the abortion rate was 77% when DS was detected using prenatal diagnostic screening [17]. In the other cases, DS was diagnosed in the first month after birth. In terms of comorbidities and intellectual deficiency, the characteristics of our population were consistent with the literature [3; 5; 18; 19].

The medical follow-up data from this study suggest that the management of children and teenagers with DS was globally satisfactory according to current recommendations (ophthalmologist, ear-nose-throat specialist, dentist, cardiologist) [6; 9; 20]. Nonetheless, the literature shows that individuals with DS who are older than 18 do not attend regular medical follow-up, despite the high frequency of medical complications in adult life [21]. This can be explained by the challenges in identifying and assessing pain in these patients but also their inability to clearly express their needs [22-23]. Furthermore, certain health professionals may lack training or knowledge about intellectual disability or the specific needs of DS patients, and there is a shortage of available health professionals and medical time.

Regarding rehabilitation, there is considerable evidence regarding the effectiveness of early intervention [18; 19; 24; 25]. An early intervention program that includes physical, occupational and speech therapy reduces parental stress and frustration [26]. It is also helpful for the overall development of the children. Unfortunately, it was not possible in our study to correlate access to early intervention and level of ID. However, our study showed that only 20% of the children attended physiotherapy consultations, which can be explained by the cessation of physiotherapy once walking is acquired, even though motor skills decrease with age [3; 24; 25]. Only a few patients attended occupational therapy sessions, but in France occupational therapy is generally limited to specific purposes, while psychomotor therapy is used for a more holistic rehabilitation, particularly in children.

Several factors can explain the observed lack of therapy in our population: the shortage of professionals in medico-social institutions, the lack of knowledge among medical doctors about the effectiveness of this type of therapy, and the non-reimbursement of private-sector occupational therapy sessions by the French health insurance (though reimbursement can be authorized by the administrative centre for disability). Our results show that one third of the patients attended follow-up exclusively in the private sector and one third used both the public and private sectors. This may reflect personal choice or the fact that it is difficult to access specialized care in the public sector, as highlighted in our qualitative study.

Regarding education, our study suggested that kindergarten was universal, and that most of the children then attended special needs classes until approximately 10 years old. Above this age, most children were relocated to a medico-social institution. These results can be explained by the French law n°2005-102. In 2005-2006, 80% of disabled children between the ages of 6 and 7 (including 72% of children with ID) were still in the mainstream school system, as compared to 52-69% between 1999 and 2005 [27,28]. The main difficulty expressed by patients and professionals was the lack of space in schools and rehabilitation facilities, especially medico-social institutions, which results in long waiting times. Moreover, the regional differences in the available health facilities may be confusing for some families [29].



From a social point of view, there was on average eight months between the diagnosis of DS and official recognition of the disability by the administrative centre for disability. This is twice more than the average time required to process regular files (3 months), but three times less than the average time needed for Prader-Willi Syndrome (22 months) [30, 31]. Nonetheless, the delay in recognition also delays the attribution of financial compensation. The length of the process may be a result of the increasing activity of these administrative centres. We also found that 5% of the patients were not receiving the financial compensation that was available to them. This may be due to parents' lack of awareness concerning their rights, their inability to clearly express their needs to the centre, or their unwillingness to make requests that would acknowledge the disability.

The time-consuming nature of the administrative procedures represented one of the main difficulties for families according to the health professionals. A study focused on the issue of waiting lists for openings in either SESSADs or in a medico-social institution was conducted in Burgundy in 2012 [32]. A "pressure" indicator, consisting of the ratio between the number of requests and the number of authorized places, was calculated. The rate was 25% for medico-social institutions and 40% for SESSADs. It appears that, despite the regular increases in the number of SESSAD places, waiting lists remain. However, due to the variability of the care offer at local level, it seems difficult to extrapolate these results on a national level.

Although our study was innovative with regard to the wealth of medico-social data provided, we acknowledge there are certain weaknesses. First, several questionnaires were not totally filled in, resulting in missing data in our data set. Moreover, the recruiting method, which was based on volunteer participation of families who were solicited by the reference centre for rare diseases or a patient support group, may have led to a selection bias. Also, the qualitative study made it possible to uncover care-related issues that could have not been obtained in a quantitative study alone (for instance, data relative to the difficulty in accessing health care were not available in the CNSA study). Finally, this study took place in the French context and so the results may not be easily generalized. Further studies are

needed in other countries.

Altogether, this study revealed ambivalent results in the medical follow-up, rehabilitation, schooling, financial and social support provided to children and teenagers with DS in France. Progress has been made especially in schooling and social aspects. Nonetheless, families still face difficulties on a day-to-day basis. It also underlined that follow-up in a reference and/or specialized centre was essential for optimising the multidisciplinary coordination of care, as indicated in the recommendations. These reference centres should ensure the continuity of care and rehabilitation into adulthood, which is part of the current public policy.

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### **Figure titles and legends:**

Figure 1. Percentage of children according to the type of rehabilitation and the age in the DS population (CNSA Study, France 2011-2012), n=82.

\*n inferior to the total participants in the study due to missing data

Figure 2. Percentage of children according to the type of schooling and the age in the DS population (CNSA Study, France 2011-2012), n=90.

\*n inferior to the total participants in the study due to missing data

### **Table titles and legends:**

Table 1. Characteristics of the participants of T21Qual study (Burgundy 2017).

\* MPDH: public administrative centre for disability

\*\* SESSAD: organisation offering rehabilitation services, educational support and home care

Table 2. Population description (CNSA Study, France 2011-2012), n=95.

Table 3. Psychomotor development of children with DS (CNSA Study, France 2011-2012), n=95.

*\*Mean normal age for: Sitting: 9 months; Standing: 12 months; Walking: 12 to 18 months; First words: 10 to 12 months; First sentence: 24 months; Toilet trained daytime: 12 to 24 months; Dry night-time: 24 to 36 months [15].*

Table 4. Medical follow-up (CNSA Study, France 2011-2012), n=92.

*\* At least one appointment once a year*

*\*\*Neuropediatrician, neuropsychiatrist, physical medicine and rehabilitation doctor, pneumologist, gastroenterologist, surgeon*

## References

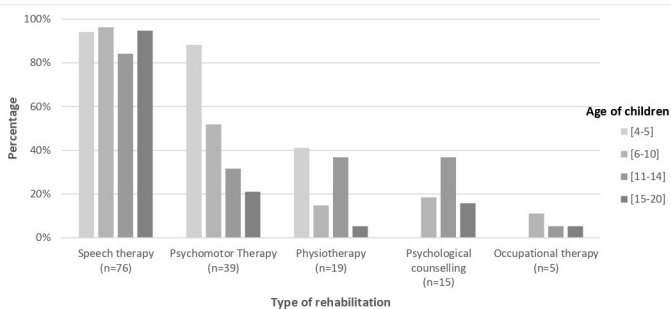
- [1] Rousseau T, Amar E, Ferdynus C, Thauvin-Robinet C, Gouyon J-B, Sagot P. Variations in the prevalence of Down's syndrome in the French population between 1978 and 2005. *J Gynécologie Obstétrique Biol Reprod.* 2010 Jun;39(4):290–6.
- [2] Rozenberg P, Bussièrès L, Senat M-V. Down syndrome screening in France : the worst consensus. *J Gynécologie Obstétrique Biol Reprod.* avr 2007;36(2):95-103.
- [3] Cuilleret M. *Trisomie et handicaps génétiques associés*. 5e éd. Abrégés. Elsevier Masson, 2007.
- [4] Loane, M., Morris, J., Addor, M. *et al.* Twenty-year trends in the prevalence of Down syndrome and other trisomies in Europe: impact of maternal age and prenatal screening. *Eur J Hum Genet* 21, 27–33 (2013).
- [5] Vicari, S. « Motor Development and Neuropsychological Patterns in Persons with Down Syndrome ». *Behavior Genetics* 36, no 3 (mai 2006): 355-64.
- [6] Fréminville B, Nivelon A, Touraine R. Suivi médical de la personne porteuse de trisomie 21, 2ème édition ; Saint-Etienne : brochure Trisomie 21 France, 2007.
- [7] Pueschel, S. M. « Clinical Aspects of Down Syndrome from Infancy to Adulthood ». *American Journal of Medical Genetics. Supplement* 7 (1990): 52-56.
- [8] France. Loi n° 2005-102 du 11 février 2005 pour l'égalité des droits et des chances, la participation et la citoyenneté des personnes handicapées. (page consulted on April 26,2016):  
<https://www.legifrance.gouv.fr/affichTexte.do?cidTexte=JORFTEXT000000809647>.
- [9] « Protocole national de diagnostic et de soins (PNDS) – Trisomie 21 ». France: Haute Autorité de Santé, janvier 2020
- [10] Makdessi Y, Mordier B. Établissements et services pour enfants et adolescents handicapés - Résultats de l'enquête ES 2010. France : Direction de la recherche, des études, de l'évaluation et des statistiques; mars 2013. 362 p. Documents de travail no. 177.

- [11] Makdessi Y. L'accueil des enfants handicapés dans les établissements et services médico-sociaux en 2010. France : Direction de la recherche, des études, de l'évaluation et des statistiques; février 2013. 6 p. Études et Résultats no. 832.
- [12] Mainguené A. Les structures pour enfants handicapés en 2006 : un développement croissant des services à domicile - Résultats de l'enquête ES 2006. France : Direction de la recherche, des études, de l'évaluation et des statistiques; novembre 2008. 8 p. Études et Résultats no. 669
- [13] O'Cathain A. Mixed methods research in the health sciences: a quiet revolution. *Journal of Mixed Methods Research* 2009;3(1):3-6.
- [14] Creswell JW, Plano Clark VL. *Designing and Conducting Mixed Methods Research*. 2nd ed. Thousand Oaks, CA: Sage; 2011.
- [15] Miles, M. B., & Huberman, A. M. (1994). *Qualitative data analysis: An expanded sourcebook* (2nd ed.). Thousand Oaks, CA, US: Sage Publications, Inc
- [16] Bourrillon A, Benoist G, Delacourt C. Item 53 : Développement psychomoteur du nourrisson et de l'enfant : aspects anormaux et pathologiques (sommeil, alimentation, contrôles sphinctériens, psychomotricité, langage, intelligence). In : *Pédiatrie*. 6<sup>e</sup> édition, Elsevier Masson, collection les référentiels des collèges, 2014, pp 59-74
- [17] « Rapport d'activité annuel du diagnostic prénatal 2016 ». France : agence de la biomédecine, avril 2017.
- [18] Roizen NJ, Patterson D. Down's syndrome. *The Lancet*. avr 2003;361(9365):1281-9.
- [19] Fidler DJ, Nadel L. Education and children with Down syndrome: Neuroscience, development, and intervention. *Ment Retard Dev Disabil Res Rev*. 2007;13(3):262-71.
- [20] Cohen WI. Health care guidelines for individuals with Down syndrome: 1999 revision. *Down Syndrome Quar* 1999; 4: 1–15.
- [21] Henderson A, Lynch SA, Wilkinson S, Hunter M: Adults with down's syndrome: The prevalence of complications and health care in the community. *Br J Gen Pract* 2007;57:50 - 55.

- [22] « Audition publique : Accès aux soins des personnes en situation de handicap ». Tome 1 Paris: HAS, 22 novembre 2008.
- [23] « Audition publique : Accès aux soins des personnes en situation de handicap ». Dossier participants. Paris: HAS, octobre 2008.
- [24] Hines S, Bennett F. Effectiveness of Early Intervention for Children with Down Syndrome. *Mental Retardation and Developmental Disabilities Research Reviews* 2, n° 2 (1996): 96-101.
- [25] Fréminville B and al. L'accompagnement des enfants porteurs de trisomie 21. *Médecine thérapeutique et Pédiatrie* 2007 ; 10(4) : 272-280.
- [26] Agarwal Gupta N, Madhulika K. « Diagnosis and Management of Down Syndrome ». *Indian Journal of Pediatrics* 81, n° 6 (juin 2014): 560-67.
- [27] De Lacerda E, Jagers C, Michaudon H, Monteil C, Tremoureux C. La scolarisation des enfants et adolescents handicapés, *Note Inf. - Dir. Program. Développement.* (n.d.) 1–10.
- [28] Espagnol P. Prouchandy P. La scolarisation des enfants et adolescents handicapés, *Note Inf. - Dir. Program. Développement.* (n.d.) 1–6.
- [29] « Accueil et accompagnement des enfants et adultes en situation de handicap ». France: CNSA, juin 2016.
- [30] Caisse nationale de solidarité pour l'autonomie (CNSA). Les MDPH face à de nouveaux enjeux-Synthèse des rapports d'activité 2014 des maisons départementales des personnes handicapées. France; Décembre 2015. 128 p. Dossier technique.
- [31] Roux-Levy P-H, Bournez M, Masurel A, Jean N, Chancenotte S, Bordes M, et al. Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. *European Journal of Medical Genetics.* déc 2020;63(12):104064.
- [32] Gérardin, Isabelle. « Etude de la liste d'attente de la MDPH 21 - Situation des jeunes inscrits en liste d'attente et état des besoins ». France: CREA Bourgogne, février

2013.

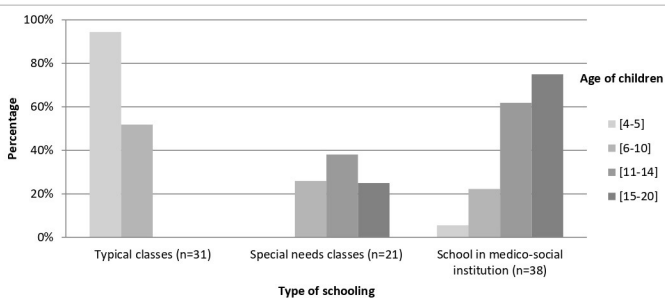
**Figure 1.** Percentage of children according to the type of rehabilitation and the age in the DS population (CNSA Study, France 2011-2012), n=82\*



\*n inferior to the total participants in the study due to missing data



**Figure 2.** Percentage of children according to the type of schooling and the age in population of children with DS (CNSA Study, France 2011-2012), n=90\*



\*n inferior to the total participants in the study due to missing data

**Table 1.** Characteristics of the participants of T21Qual study (Burgundy 2017)

<b>Participant</b>	<b>Sex</b>	<b>Job</b>	<b>Employer</b>
P1	F	Medical doctor	Medico-social institution
P2	F	Social worker	MDPH*
P3	F	Medical doctor	MDPH
P4	F	President	Down syndrome association
P5	F	Speech therapist	Private sector
P6	F	Director	SESSAD**
P7	F	Medical doctor	SESSAD
P8	M	Special needs teacher	SESSAD
P9	F	Speech therapist	SESSAD
P10	F	Special needs teacher	SESSAD
P11	F	Medical doctor	SESSAD
P12	F	Project manager	Regional Health Agency
P13	F	Managing director	Regional Health Agency

\* MPDH: public administrative centre for disability

\*\* SESSAD: organisation offering rehabilitation services, educational support and home care

**Table 2.** Population description (CNSA Study, France 2011-2012), n=95

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<b>Age</b> (n=91, years) (mean,SD)	10.9 (4.8)
<b>Sex (n,%)</b>	
Male	54 (56.8)
Female	41 (43.2)
<b>Marital status of the biological parents (n,%)</b>	
Living as couple	71 (74.7)
Separated	15 (15.8)
Adopted child	8 (8.4)
Foster family	1 (1.1)
<b>Number of additional siblings (n,%)</b>	
0	9 (9.5)
1	26 (27.4)
2	21 (22.1)
≥3	31 (32.6)
Missing Data	8 (8.4)
<b>Intellectual disability (n,%)</b>	
Learning Disability	1 (1.1)
Mild	5 (5.3)
Moderate	63 (66.3)
Severe	18 (18.9)
Profound	5 (5.3)
Missing Data	3 (3.2)
<b>Antenatal diagnosis (n,%)</b>	
No	77 (81.1)
Yes	12 (12.6)
Missing data	6 (6.3)

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**Table 3.** Psychomotor development of children with DS (CNSA Study, France 2011-2012), n=95

	Mean age of acquisition in months (SD)*
Sitting	10.9 (3.9)
Standing	18.9 (7.0)
Walking	27.5 (8.8)
First words	29.0 (14.4)
First sentence	56.7 (26.5)
Toilet trained daytime	50.0 (27.4)
Dry night-time	67.0 (33.5)

\*Mean normal age for: Sitting: 9 months; Standing: 12 months; Walking: 12 to 18 months; First words: 10 to 12 months; First sentence: 24 months; Toilet trained daytime: 12 to 24 months; Dry night-time: 24 to 36 months [16].

**Table 4.** Medical follow-up\* (CNSA Study, France 2011-2012), n=92

	Number of patients (%)
Specialists involved	
Ophthalmologist	80 (87.0)
General practitioner	77 (83.7)
Dentist	65 (70.7)
Ear-nose-throat specialist	64 (69.6)
Medical geneticist	38 (41.3)
Cardiologist	35 (38.0)
Orthopaedist	29 (31.5)
General Paediatrician (private practice)	22 (23.9)
General Paediatrician (hospital)	19 (20.7)
Dermatologist	15 (16.5)
Endocrinologist	8 (8.7)
Others**	35 (38.0)

\*At least one appointment per year

\*\*Neuropediatrician, neuropsychiatrist, physical medicine and rehabilitation physician, pneumologist, gastroenterologist, surgeon