Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome


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Title page

Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome.

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The authors declare no conflict of interest

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Abstract

Prader-Willi syndrome (PWS) is a rare genetic neurodevelopmental disorder with a characteristic behavioural phenotype. A multidisciplinary approach to care is required to prevent multiple medical complications in individuals affected by PWS. The aim of this study was to describe the rehabilitation, medical care, educational and social support provided to school-aged French PWS patients with varying neuropsychological profiles. Data were obtained from a French multicentre study that included patients aged 4 to 20 years with diverse genetic syndromes. Nineteen PWS subjects with a mean age of 9.2 years were included. The mean full-scale intellectual quotient (IQ) was 58 (Wechsler scale). There were frequent dissociations between verbal and performance IQ that were not associated with a specific profile. We also observed lower autonomy and communication scores (5.3 years and 5.9 years equivalent, respectively, Vineland scale), the absence of hyperactivity (Conners scale), and the presence of behavioural abnormalities (CBCL scale). Multidisciplinary medical supervision was generally coordinated by the paediatric endocrinologist and did not always include follow-up with all of the recommended specialists, in particular with a paediatric psychiatrist. Analysis of multidisciplinary rehabilitation conducted in public and private-sector establishment revealed failings in psychological support, occupational therapy and dietary follow-up. Regarding education, most children younger than 10 years were in normal schools, while older individuals were often cared for in medico-social institutions. In conclusion, children and adolescents with PWS generally received appropriate care. Though there have been considerable improvements in the management of children with PWS, reference centres should continue reinforcing the coordination of multidisciplinary supervision.

Key words: Prader-Willi syndrome; intellectual disability; patient care management; social support
Main Text

Introduction

Prader-Willi (PWS) syndrome is secondary to abnormal expression of genes within a region subject to parent-specific imprinting on chromosome 15. It affects about 1 in 20,000 births and is characterized by severe hypotonia and feeding difficulties in early infancy followed by the absence of satiety and hyperphagia in early childhood [1]. Without multidisciplinary care, children with PWS will become morbidly obese [2–6]. Motor milestones and language development are delayed and often aggravated further by psychological and behavioural issues. All individuals have some degree of cognitive impairment, but the degree varies widely from one child to another. A distinctive behavioural phenotype (temper tantrums, stubbornness, manipulative behaviour, and obsessive-compulsive characteristics) is common, as are physical attributes such as short stature, characteristic facial features and small hands and feet. Comorbidities include endocrine dysfunction such as growth hormone deficiency, hypothyroidism, hypogonadism in both males and females with genital hypoplasia, incomplete pubertal development, and infertility. Strabismus, scoliosis, osteoporosis, skin picking, behaviour problems, impaired emotional skills, poor social abilities and psychiatric disorders are also frequently described [7,8].

In infancy, management includes nasogastric tube feeding and monitoring of oral skills to assure adequate nutrition, physical therapy to improve muscle strength, and hormone substitution therapy with surgical treatment for cryptorchidism. The major challenge in childhood is regulating weight gain. The early introduction of growth hormone replacement therapy can dramatically improve the health of PWS patients as it helps to normalize height, increase lean body mass and mobility and decrease fat mass, especially when accompanied by strict control of access to food and the monitoring of height, weight, and body mass index [9–21]. In adulthood, growth hormone therapy may be continued to maintain muscle mass,
but there are no data in terms on the long-term effect of treatment in adults, particularly concerning the effect on behavioural problems and autonomy [22–27].

Other issues include the evaluation and treatment of sleep disturbances, sex hormone replacement therapy at puberty, rehabilitation and the treatment of behavioural problems. Rehabilitation can include speech therapy, physiotherapy, occupational therapy, psychomotor therapy, and psychological support. In adulthood, the regulation of behaviour and weight management, including physical activity to prevent morbid obesity, remains the major challenge.

Therefore, management should be global, multidisciplinary and focused on rehabilitation, medical care, and social support, and ideally be coordinated by one institution. Between 2004 and 2007, reference centres for rare diseases received accreditation for the French Ministry of Health for this type of coordination. Additional aid can be provided by specific patient support groups whose purpose is to help affected individuals and their families obtain support and recognition (Prader-Willi France is the main PWS association in France).

In France, law n°2005-102 of the 11th February 2005, provides guidelines for the care of disabled persons and has led to the creation of a local government agency for disabilities. These local agencies set the financial compensation needed to pay for care, technical equipment, and special needs in schooling. Rehabilitation can be offered at the child’s home, at school, or in a private centre through the department of special education and home care, otherwise it takes place in a medico-social institution. Children with a cognitive disability but no intellectual disability (ID) can attend regular school, often with the support of an educational assistant, or an integrated school with fewer children, specialised teachers and adapted teaching materials. Individuals with ID attend special schools for patients with ID (in medico-social institution), that combine social support, rehabilitation and medical care.

Law n°2005-102 was a clear step towards improving the situation of disabled persons in France, but families still often find it challenging to obtain medical care, social and
educational support, and appropriate rehabilitation. In this context, using data from a French national study, we aimed to describe the medical care, rehabilitation, and social and educational support provided to children with PWS relative to their degree of ID.

**Material and methods**

*The national CNSA study*

The National Solidarity Fund for Autonomy (*Caisse Nationale de Solidarité pour l’Autonomie* - CNSA) ordered and financed a study of the quality of care provided to school-aged patients with chromosomal abnormalities and intellectual disabilities as well as their cognitive and behavioral level. The aim was to pinpoint the existing qualitative and quantitative deficiencies in the social, educational and medical support provided to these children. Nine French reference centres for rare diseases were enrolled in the study. The inclusion period went from mid-2011 to the end of 2012. The inclusion criteria were: children between 4 and 20 years with a proven chromosomal abnormality (standard karyotyping, FISH or array-CGH), National Health Insurance Agency coverage, and written informed consent for enrolment in the study. Patients under guardianship or in juvenile detention were excluded. The reference centres or patient associations contacted parents by postal letter or phone to take part in the study. 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, age, weight, height, etc.), demographics (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 social benefits, parental work, etc.) and satisfaction. Parents and children were then asked to spend one day at their
reference centre. During the visit, the questionnaire was verified with the help of an in-house investigator.

Then, a neuropsychologist recruited for the study conducted a neuropsychological evaluation of the child using several scales: i) Wechsler’s scales (according to age, WPPSI-III, WISC-IV or WAIS-III), used for an overall assessment of intellectual performance comprising four sub-domains (verbal, non-verbal, working memory and processing speed) [28,29]; ii) the Vineland Adaptive Behavior Scales-II, with investigations in four domains (autonomy, communication, socialization and motor function) to provide a better view of patient autonomy than IQ [30]; iii) the Vineland Yields standard scores and the Child Behavior Checklist (CBCL) to describe patient behaviour and personal or social self-sufficiency [31]; and iv) the Conners Parent Rating Scale (CPRS) and Conners Teacher Rating Scale (CTRS) to identify children with disruptive behaviours, in particular attention disorders and hyperactivity [32,33]. For the purpose of clarity, regardless the type of scale, PIQ will be the abbreviation used for the performance indices, VIQ for the verbal indices and TIQ (total IQ) throughout the manuscript.

Statistical studies

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 exact 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 9.3.

Results

Population description
The characteristics of the 19 patients included in the study are presented in Table 1. All patients had PWS secondary to deletion. Our cohort were 58% female and 42% male, and 74% were living in an urban area. They originated from various socio-economic backgrounds (results not shown). PWS was diagnosed following neonatal hypotonia (93% of the cases), neonatal feeding issues (21%) or other neonatal problems (11%). Though the definitive genetic diagnosis was obtained at a mean age of 10.4 months (SD = 17.5), 76.5% of the patients were diagnosed before the age of 5 months. The remaining individuals were diagnosed after the age of 30 months, and half were 17 years old at diagnosis. Psychomotor development is described in Table 2. There was a global delay in all stages of psychomotor development with varying degrees of severity.

Neuropsychological and behavioural evaluations

Figure 1 shows the distribution of TIQ, VIQ, and PIQ. The mean TIQ was 57.9 (SD = 13.4), with slight variations depending on the test. No specific neuropsychological profile was found. Interestingly, one 6.2-year-old patient had no intellectual disability (TIQ = 83) and three had a borderline TIQ (70 < TIQ < 80). The PIQ was greater than the VIQ in 59% of cases, and the VIQ was greater than the PIQ in others.

Developmental delays were confirmed with the Vineland scale, which resulted in mean age equivalents of 5.3 years for communication (SD = 3.7), 5.9 years for autonomy (SD = 3.7), 6.0 years for socialization (SD = 2.2), and 4.1 years for motor skills (SD = 1.0). The parents’ and teachers’ responses for the Conners scale did not reveal any particular behavioural disorders. The CBCL scale for the internalizing domain showed a pathological score in 56% of the children, a borderline score in 28% and a normal score in 17%. In the externalizing domain, 28% of children had a pathological score, 11% a borderline score and 61% a normal score.

Medical care
Medical monitoring was often multidisciplinary and coordinated by a general practitioner or a paediatric endocrinologist in 95% and 89% of cases, respectively (Table 3). The average number of medical consultations (specialized or not) was 12.5 per year. Patients were treated with growth hormone in 79% of the cases and L-thyroxine in 58%. At the time of the study, the patients who were not receiving growth hormone therapy were between 6 and 17 years.

Rehabilitation

The children had an average of three types of rehabilitation. Speech therapy, physiotherapy and psychomotor therapy were the most frequent type of rehabilitation (Table 4). For 65% of the children, most rehabilitation sessions took place in public-sector social-educational institutions; the remaining 35% of sessions were in the private sector. However, it varied according to the type of rehabilitation: private-sector care was most frequent for physiotherapy and speech therapy (92% and 67%, respectively), while psychomotor therapy and psychological support were most frequently in public-sector establishments (100% and 80%, respectively). The average number of rehabilitation sessions per child was 3.2 per week. The mean TIQ did not significantly differ with private or public-sector rehabilitation.

Education

The type of educational facility is summarized in Table 5, according to the age and cognitive level of each child. One patient was home schooled with correspondence courses. Most of the patients aged less than 10 years were in normal schools and most of the older patients were in medico-social institutions. The mean time spent at school was 26 hours per week. An educational assistant was present for all of the children in a normal school. The waiting time for the attribution of an educational assistant was a mean of 3.9 months after the request was submitted (SD = 3.2 months).

The type of schooling was not significantly linked to the mean TIQ, but was mainly associated with the patient’s age. No significant difference was found between the mean
scores of the Vineland for communication, autonomy, socialization and motor skills of children and adolescents in normal schools compared with those of children in medico-social institutions. The parents attributed significantly better scores on the Conner's scale than teachers, in particular for total hyperactivity (p = 0.03), behavior (p <0.001) and impulsivity / hyperactivity (p <0.001).

Social

The average time between the diagnosis of PWS and recognition of the disability by a local agency was 22 months (SD= 29) (Figure 2). Ten of the mothers indicated that they had gone from a full-time job to a part-time job because of their child's disability. For 70% of the mothers, the main reason for this change was to be able to attend their child's regular appointments for medical care and rehabilitation. In contrast, two fathers changed from full-time to part-time work, and only one because of his child's disability. Forty-two percent of the parents reported that the grandparents provided caregiving on a regular basis. Forty-two percent also reported the existence of global constraints, and 37.5% had changed their primary residence because of their child's disability. One quarter of the parents reported making adjustments in their home in order to meet the specific needs of their child.

Discussion

The present study is the first to use data from a national study to analyse the medical care, rehabilitation, and social and educational support received by patients with PWS in relation to their degree of intellectual disability (ID). Our study is also novel in that it is the first to use such comprehensive medico-social data, but the cohort of patients was small (19 children) and some data was missing. Moreover, there was a potential selection bias due to the recruiting method that was based on volunteer families contacted by the reference centre for rare diseases or through patient support groups.
The design of this study revealed how and when the children were diagnosed. The mean age at diagnosis was later than that previously reported in a larger French cohort, which was 2 months [35]. Our later mean can be explained by the inclusion of older children who were born before the PWS awareness campaign for paediatricians underlining the need to screen in cases of neonatal hypotonia. The medical follow-up data from this study suggest that the management of PWS children and teenagers was globally satisfactory, and that regular multidisciplinary care was provided in accordance with the French national protocol for diagnosis and care [36]. Indeed, 95% of the patients were followed by an paediatric endocrinologist or an endocrinologist, and three quarters of the patients were treated with growth hormones, which have proven to be effective in this syndrome [13,14,21,22]. Follow-up with an ophthalmologist, ear-nose-throat specialist, orthopaedic specialist and dentist were regularly implemented for most patients, but the annual frequency was lower than recommended. In addition, follow-up with a paediatric psychiatrist was insufficient (only 11% patients had regular consultations) even though such care is strongly recommended. It was not possible to determine whether the lack of follow-up was a result of limited accessibility or awareness, problems with referrals, family choice or care overload. Finally, we found that dietary follow-up was insufficient (5%), even though it is particularly important in PWS (Table 2).

Most of the patients in this study had mild or moderate ID, which is concordant with the literature [37–40]. The cognitive profile was heterogeneous (higher PIQ or higher VIQ), which is difficult to explain. However, according to the literature, there was a trend towards equivalent VIQ and PIQ and better visuo-spatial skills in PWS secondary to deletion. On the contrary, better verbal skills than non-verbal skills were observed in PWS secondary to uniparental disomy (the difference often being related to praxis disorders and impaired visuo-spatial skills) [38,39]. Therefore, the fact that our study only included patients with PWS secondary to deletion could have resulted in a selection bias.

Speech therapy, physiotherapy and psychomotor therapy were initiated relatively
early in most patients, though there were a few for whom it was much later. Only a few patients had occupational therapy though it is essential for improving handwriting skills, bimanual coordination, visuomotor skills and motor control (as a complement to psychomotor therapy) which are all fundamental skills for school. There was a distinct lack of psychological management, which may be the result of several factors: the shortage of occupational therapists in medico-social establishments, the lack of knowledge among professionals about the effectiveness of this type of therapy, the fact that French health insurance does not reimburse private-sector occupational therapy sessions though they can be authorized by the local government office. One third of the patients attended exclusive private-sector facilities for rehabilitation. This could reflect parent choice or difficulties accessing public services. The use of private or public sector rehabilitation services did not seem to be related to the family’s primary residence (urban or rural).

With regard to education, schooling in normal or integrated schools was the rule for children under 10 years old, with a satisfactory number of hours of instruction and the systematic presence of an educational assistant. Above this age, most children were relocated to a medico-social establishment. These results have been influenced by the provisions of law n°2005-102. Indeed, 80% of disabled children between the ages of 6 and 7 (including 72% of children with ID) were still in the normal school system in the 2005-2006 period, as compared with 52-69% between 1999 and 2005 [41,42].

It was interesting to analyse the associations between schooling and Vineland scale scores. The Vineland scale revealed an acquisition gap, especially for communication, socialization and autonomy, which underlines the need for personalized rehabilitation and appropriate teaching for schooling to be effective.

From a social point of view, the time lapse between the diagnosis of PWS and recognition of the disability still seemed to be long. On average, the disability was recognized by the government one year after the diagnosis, thus delaying the attribution financial compensation by the same amount of time. The length of the process may be due to parents’
lack of awareness concerning their rights, their inability to clearly express their needs to the local government agency or their reticence to make requests that would acknowledge the disability.

Altogether, this study revealed mixed results in the medical follow-up, rehabilitation, schooling and social support provided to children and teenagers with PWS in France. Our results highlighted the fact that an early and accurate diagnosis resulted in earlier official recognition and the financial and social support needed to deal with the everyday difficulties linked to the syndrome. 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 must ensure the continuity of care and rehabilitation into adulthood.

Acknowledgements

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Figures titles and legends:

Figure 1. Distribution of total intelligence quotient, verbal intelligence quotient and performance intelligence quotient in the PWS population.

Regardless the type of scale (WPPSI, WISC, WAIS), PIQ was used as the abbreviation used for the performance indices, VIQ for the verbal indices and TIQ for the total index.

An intellectual disability is defined by a TIQ score under 70. A profound intellectual disability (ID) correspond with a TIQ score under 19, a severe ID with a TIQ between 20-34, a moderate ID with a TIQ between 35-49, a mild ID with a TIQ between 50-69.

Figure 2. Delay between age (months) for diagnosis and recognition of the disability.

Patients were sorted in ascending order from the youngest to the oldest.

Tables titles and legends:

Table 1: Population characteristics

*Missing data

Table 2: Psychomotor development

*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 nighttime: 24 to 36 months.

**SD: standard deviation

Table 3: Medical follow-up

Table 4: Type, location and patient age at initiation of rehabilitation and educational support.
*Public sector: department of special education and home care and medico-educational institutions

** Missing data

NA: information not available

Table 5: Education type, with intellectual level and patient age

*ID: Intellectual disability

**Education assistant only justified in case of normal school or integrated school

***NA: Not available
References


[18] L.A. Gondoni, L. Vismara, P. Marzullo, R. Vettor, A. Liuzzi, G. Grugni, Growth hormone therapy improves exercise capacity in adult patients with Prader-Willi syndrome, J.


Figure 1. Distribution of total intelligence quotient, verbal intelligence quotient and performance intelligence quotient in the PWS population.

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Figure 2. Elapsed time between diagnosis and recognition of the disability (in months)

Patients were sorted in ascending order from the youngest to the oldest.
Figure 1: perioperative protocol

Urgent surgery

Sars-CoV2 detection with RT-PCR and chest CT

Surgery in dedicated COVID+ room
Extubation, wakening and monitoring until test results are available

COVID positive

Intensive care unit COVID+
Surgical unit COVID+

COVID negative

Intensive care unit COVID-
Surgical unit COVID-

In-hospital contamination
Table 2. Psychomotor development (n=19)

<table>
<thead>
<tr>
<th></th>
<th>Mean age of acquisition (months)*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sitting</td>
<td>11.0 (SD** = 4.9)</td>
</tr>
<tr>
<td>Standing</td>
<td>21.3 (SD** = 12.4)</td>
</tr>
<tr>
<td>Walking</td>
<td>26.0 (SD** = 11.5)</td>
</tr>
<tr>
<td>First words</td>
<td>22.5 (SD** = 16.0)</td>
</tr>
<tr>
<td>First sentence</td>
<td>39.9 (SD** = 12.6)</td>
</tr>
<tr>
<td>Toilet trained daytime</td>
<td>34.7 (SD** = 8.4)</td>
</tr>
<tr>
<td>Dry nighttime</td>
<td>58.5 (SD** = 29.1)</td>
</tr>
</tbody>
</table>

*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 nighttime: 24 to 36 months [34].

**SD: standard deviation
<table>
<thead>
<tr>
<th>Medical follow-up</th>
<th>Number of patients (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Specialists involved</strong></td>
<td></td>
</tr>
<tr>
<td>paediatric endocrinologist</td>
<td>18 (95)</td>
</tr>
<tr>
<td>general practitioner</td>
<td>17 (89)</td>
</tr>
<tr>
<td>ophthalmologist</td>
<td>15 (79)</td>
</tr>
<tr>
<td>dentist</td>
<td>14 (74)</td>
</tr>
<tr>
<td>general paediatrician</td>
<td>10 (53)</td>
</tr>
<tr>
<td>orthopaedist</td>
<td>10 (53)</td>
</tr>
<tr>
<td>ear-nose-throat specialist</td>
<td>8 (42)</td>
</tr>
<tr>
<td>medical geneticist</td>
<td>7 (37)</td>
</tr>
<tr>
<td>paediatric neurologist</td>
<td>4 (21)</td>
</tr>
<tr>
<td>pneumologist</td>
<td>4 (21)</td>
</tr>
<tr>
<td>paediatric psychiatrist</td>
<td>2 (11)</td>
</tr>
<tr>
<td>visceral surgeon</td>
<td>0 (5)</td>
</tr>
<tr>
<td>gastroenterologist</td>
<td>0 (5)</td>
</tr>
<tr>
<td><strong>Medication</strong></td>
<td></td>
</tr>
<tr>
<td>growth hormone</td>
<td>15 (79)</td>
</tr>
<tr>
<td>L-thyroxin</td>
<td>11 (58)</td>
</tr>
</tbody>
</table>
## Table 4. Type, location and patient age at initiation of rehabilitation and educational support (n=19)

<table>
<thead>
<tr>
<th>Service</th>
<th>Number of patients (%)</th>
<th>Rehabilitation started before 12 months in percent**</th>
<th>Mean age at initiation in months (SD)**</th>
<th>Number of rehabilitation activities performed in private sector**</th>
<th>Number of rehabilitation activities performed in public sector***</th>
<th>Rehabilitation activities requested by the patient but not provided**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Speech therapy</td>
<td>15 (79%)</td>
<td>33%</td>
<td>27.1 (23.1)</td>
<td>10</td>
<td>5</td>
<td>0</td>
</tr>
<tr>
<td>Physiotherapy</td>
<td>13 (68%)</td>
<td>92%</td>
<td>14.9 (49.6)</td>
<td>11</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Psychomotricity</td>
<td>12 (63%)</td>
<td>50%</td>
<td>24.5 (27.0)</td>
<td>2</td>
<td>8</td>
<td>0</td>
</tr>
<tr>
<td>Psychology</td>
<td>8 (42%)</td>
<td>50%</td>
<td>34.5 (38.6)</td>
<td>0</td>
<td>5</td>
<td>3</td>
</tr>
<tr>
<td>Occupational therapy</td>
<td>4 (21%)</td>
<td>75%</td>
<td>9.0 (18.0)</td>
<td>1</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>Orthoptics</td>
<td>4 (21%)</td>
<td>50%</td>
<td>28.5 (33.0)</td>
<td>3</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Dietary</td>
<td>1 (5%)</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
</tr>
</tbody>
</table>

* Public sector: department of special education and home care and medico-educational institutions  
** Missing data  
NA: information not available
<table>
<thead>
<tr>
<th>Age (years)</th>
<th>Level of ID*</th>
<th>School type</th>
<th>Educational assistant (AVS)**</th>
<th>Instruction time at regular school</th>
</tr>
</thead>
<tbody>
<tr>
<td>4</td>
<td>Mild</td>
<td>Normal school</td>
<td>+</td>
<td>29 hours (8 half-days)</td>
</tr>
<tr>
<td>4</td>
<td>None</td>
<td>Normal school</td>
<td>+</td>
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</table>

*ID: Intellectual disability

**Education assistant only justified in case of normal school or integrated school

***NA : Not available