The Care Pathway for Trisomy 21 Cases in Algeria: Between Health and Social Care

Le parcours de soins des cas de trisomie 21 en Algérie : entre santé et protection sociale

Sarra Samra Benharrats [1,*]
[1] Sidi Chami Psychiatric Hospital of Oran, Faculty of Medicine,
University of Oran 1 – Ahmed Ben Bella, Algeria

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Abstract: Trisomy 21 or Down syndrome is a relatively common disability known to health professionals and Algerian society in general. This type of disability requires special follow-up and specialized care, which is mainly based on the screening of the disability in the first place and its secondary somatic and psychic complications which manifest themselves throughout the life of Down's syndrome from birth to senescence. This type of screening requires the involvement of parents and specialized resource persons in particular health professionals. However, its success depends on several factors, both social and health, which can guarantee the prevention of any complication and above all improvement of the quality of life of subjects with this type of disability. In this work, we present a relationship decryption of children's parents with Down's syndrome with the health system and the vagaries of their care pathway in the screening of somatic and psychic disorders related to Down's syndrome, emphasizing the unavoidable interest of early detection to overcome all difficulties encountered during the life of the subject with trisomy 21.

Keywords: Trisomy 21; Disability; Screening; Care; Complications.

الملخص : يعتبر التثلث الصبغي 21 أو ما يُعرف بمتلازمة داون من الإعاقات الشائعة نسبيًا لدى المهنيين الصحيين والمجتمع الجزائري بشكل عام. هذا النوع من الإعاقة يتطلب مراقبة خاصة ورعاية متخصصة، والتي تقوم بالدرجة الأولى على فحص الإعاقة أولاً والمضاعفات الجسدية والنفسية الثانوية التي تظهر طوال حياة الشخص المصاب بمتلازمة داون من الولادة وحتى الشيخوخة. يتطلب هذا النوع من الفحص مشاركة الآباء والأشخاص ذوي الخبرة المتخصصين في الموضوع، ولا سيما المهنيين الصحيين. ومع ذلك، فإن نجاحها يعتمد على عدة عوامل اجتماعية وصحية، والتي يمكن أن تضمن الوقاية من أي مضاعفات وخاصة تحسين نوعية حياة الأشخاص الذين يعانون من هذا النوع من الإعاقة. نقدم في هذا العمل دراسة وتحليل لعلاقة أولياء أمور الأطفال المصابين بمتلازمة داون بالنظام الصحي ومخاطر مسار رعايتهم في فحص الاضطرابات الجسدية والنفسية المتعلقة بمتلازمة داون، مع التأكيد على الاهتمام الذي لا مفر منه بالاكتشاف المبكر للتغلب على كل الصعوبات خلال حياة المصاب بمتلازمة داون.

الكلمات المفتاحية : التثلث الصبغي 21 ;الإعاقة; الكشف المبكر; العلاج; المضاعفات.

* Corresponding author, e-mail: benharrats.sarra@univ-oran1.dz.

1- Introduction

According to the Algerian National Statistics Office, there are two million people with disabilities, including 167,331 mentally disabled (Moudjahid, 2010). The types of these mental disabilities are numerous and diverse, including trisomy 21 or so-called Down syndrome regarding John Langdon-Down (Dalla Piazza & Dan, 2001) (Down, 1866). It is a congenital malformation expressed by the existence of a supernumerary chromosome: a polyploidy of chromosome 21, often of maternal origin, and this in about 95% of cases. People with Down syndrome have 47 chromosomes instead of the normal 46. This disability is referred to as an intellectual disability of varying degrees, ranging from mild to profound (Rondal, 2010). This type of chromosomal abnormality is responsible for about 28% of cases of mental retardation (Curry, et al., 1997).

Its overall incidence is estimated at 1/750 live births, and the risk increases gradually with maternal age (Dolk, et al., 1990). Clinically, the subject carrying this syndrome is distinguished by his physiognomnia (Verloes, 2015) (Larouse médical, 2002), consisting of a constellation of mental, physical, and functional abnormalities resulting from the triplication of chromosome 21 during meiosis (Futura-Santé, 2021). Specialists recognize three forms of trisomy 21 according to the degree of damage to organic cells, with no difference in the degree of cognitive and language impairment (Comblain & Thibaut, 2009). We note that: the standard form represents 95% of cases; the mosaic form 2% of cases; the translocation form has a prevalence of 3% of cases.

The organic and mental complications encountered in the subject with Down syndrome are numerous and can be life-threatening. They affect different organs, such as the cardiovascular system (intraventricular communication), the endocrine system (diabetes, hypothyroidism), the gastroenterological system (Hirschsprung's disease and celiac disease), the nervous and psychological system (epilepsy, muscle hypotonia, sleep disorders, depression, behavioral disorders) (Vaessen, et al., 2012). Despite the seriousness of its complications and their evolutions which can be fatal in relation to various somatic and mental impairments. The early detection of the latter remains under the influence of a prudishness of investigation in response to multiple causes, both in relation to the lack of information on the situation of the handicap by the entourage of the Down's syndrome and especially the lack of training of the specialists who take care of them. This makes the care of the child with Down syndrome difficult and perilous, both from a health and social point of view.

In this article, we illustrate the health journey of children with Down syndrome with the health system and vagaries in the process of screening for somatic and psychic disorders related to disability. We insist on the methods and interest of screening for trisomy 21 by approaching the different phases of the life of the subject with trisomy ranging from birth to senescence, by evoking the difficulties encountered in the entourage during care, and the risks incurred by this syndrome in the absence of investigation and adequate care in terms of time and quality.

2- Early Screening of Trisomy 21

Mutual intervention by the specialist and the parents of children with Down syndrome in screening is crucial in the early phases of the life of the subject with Down syndrome. Parents have difficulty detecting this syndrome in their children, especially the physiognomonic signs, because some show no unusual physical characteristics at birth and then develop characteristic facial dysmorphism during infancy. Affected newborns tend to be apathetic, rarely cry, and exhibit muscle hypotonia, these signs are seen by parents as positive elements, where the child is considered a calm baby. It is only in early childhood that the signs become clearer to observe and parental concern arises.

Whatever the period of diagnosis of this disability, it is important to have multidisciplinary support for the parents and the child by health professionals and social professionals specializing in disability, in particular trisomy 21. The prenatal period is an essential phase for screening for this type of syndrome. The diagnosis is possible by measuring the nuchal translucency, which is a space located at the neck of the fetus, carried out during a simple ultrasound by a gynecologist, or the dosage of serum markers, substances secreted by the plasma or blood test of the fetus. Moreover, the possibility of using amniocentesis allows a prenatal diagnosis with certainty.

These techniques are possible to perform during any pregnancy at risk of having a child with Down syndrome, referring to the statistical probabilities mentioned above. Nevertheless, their application is very modest, to the point that some practitioners prefer not to investigate and say nothing to future parents even if they notice an anomaly for fear of alarming them. The practitioners justify this by the fact that even if the diagnosis is made, the pregnancy will not be terminated. This vision is correct but incomplete, because it is not about the question of termination of pregnancy but about the preparation of future parents to take charge of this type of disability in their children. A preparation that includes the psychological support of these parents and especially training them to better understand and adapt to this new situation that affects their offspring.

Leaving future parents of children with Down syndrome to their fate and far from any understanding of this handicap is a major risk for the child and for the whole family. Indeed, parents can develop a psychological shock at the sudden discovery of their child's disability, thus causing a major risk in terms of its acceptance and care, by switching between denial and rejection of the disability, or even of their child. After the birth of the child with Down syndrome, parents often find themselves on their own, rarely are the advice and recommendations given from the first day of birth.

At this stage, parents must know the importance of screening for somatic and mental pathologies, and especially the interest of their early treatment to improve the vital prognosis of their child. Comorbid organic and mental pathologies are numerous and can be fatal in the long term through the risk of decompensation. In this phase precisely, the parents report their psychological suffering linked to the mourning of the “normality” of their child. Furthermore, their sufferings related to their introduction in a bureaucratic spiral to prove the handicap of this child with Down syndrome. These same parents are forced to perform a karyotype in centers specializing in genetics to regularize the disability file which entitles them to free care.

This file contains several administrative and medical documents which are most often renewed annually. In this context, parents of children with Down's syndrome and even professionals, in particular psychiatrists who have to write this type of report, complain about this inconsistency, because each year they have to confirm the presence of Down's syndrome in these children, as if it were a reversible condition and not a permanent handicap.

Adding to this, another difficulty linked to the incomplete and reduced training of certain practitioners vis-à-vis this disability, making care difficult for these children, once again introducing families into another medical spiral. On this subject, some practitioners believe that it is not a question of a real handicap but of a unique physiognomy of its kind, thus neglecting the possibility of any probable complication. To remedy this situation, it is important to note the primacy of coordination between the family, medical caregivers and social workers, or even charities for disabled children, for the supervision and care of parents and children with Down syndrome.

3- The Down Syndrome Care Process

The evolution of somatic and mental clinical signs in children with Down syndrome is variable and depends on the form of disability and the degree of early intervention by parents and specialists. This intervention itself depends on the age of the child with Down syndrome and its stage of diagnosis. It extends from the prenatal period to birth and then the process continues until adulthood, and even beyond, and this, according to its life expectancy. In the prenatal phase, it is important to insist on the training of professional health specialists to diagnose this type of disability in time and to raise awareness among future parents who meet

the probability of risk factors for having a child with Down syndrome on this congenital pathology and train them on its clinical and behavioral signs. Additionally, support these parents both on a medico-psychological and social level throughout this process.

The prenatal phase is followed by a birth phase which concerns the announcement of the diagnosis of trisomy 21 to the parents, which is the first therapeutic and support act for the parents, and this, by detecting the physiognomic signs of the newborn. Data from studies of the neuropathology of children with Down syndrome suggest that the main differences between normally developing children and children with Down syndrome only emerge a few months after birth (Ecalle & Magnan, 2002), sometimes making the acceptance of the diagnosis by the parents of these children difficult or even impossible. As soon as the diagnosis is established, it is important to inform the parents about the progressive risks of this syndrome. Thus, they must be informed about probable pathologies and secondary complications. It is also important to inform them and direct them to resource people to help them take care of their child to cushion this type of complication and above all to help them improve their quality of life through training and education and adapted learning to combat cognitive decline. This phase is crucial for the medico-psychological support of the parents and the child where organic comorbidities can be detected. It is considered as a phase of prevention and diagnosis of pathologies secondary to Down syndrome.

Early support is offered since the first months, i.e. between 3 and 6 months to promote the proper development of the neuromotor schema, motor skills, communication and language, body schema and perception, as well as interaction with the environment. The early childhood of Down syndrome is usually characterized by the risk of psychomotor retardation and mental retardation. The child with Down syndrome walks between 2 and 3 years old and speaks around 4 to 5 years old. On average, he has an intelligence quotient (IQ) estimated at 50, but thanks to good educational support, and early and targeted school learning, this IQ can be greatly increased for the benefit of this child. The most significant deficits can only be observed beyond the first year of life in the learning of very specific information such as places or certain aspects of language. Some deficits are more abstract in nature and result in instability of learned material, where information tends to be forgotten soon after it is learned (Ecalle & Magnan, 2002).

To ensure better learning for children with Down syndrome, special classes have been designed. They are mainly located in the big cities of Algeria, such as Algiers, Oran, Annaba, and Tlemcen. These special classes can be beneficial for promoting targeted and personalized school supervision for children with Down syndrome, except that emulation in the school

environment with other normal children can be more beneficial for the learning of children with this handicap, and this, by behavioral and cognitive contagion effects.

Unfortunately, this type of special classes for children with Down syndrome are rare and non-existent in several regions of the national territory, we cite the highlands and southern Algeria, precisely in rural areas. It should be noted that these areas may contain a large population of people with trisomy 21. The statistics do not inform us about this detail, but we can consider that it is very probable that there is a significant prevalence of this handicap, and this, because of the maintenance of the desire to give birth despite the advanced age of the mother, who most often is multiparous. Without omitting another risk factor for this handicap, which is consanguineous marriage, which is very common in the rural areas of the aforementioned areas.

Finally, during the phase that extends from adolescence to adulthood, the subject with Down syndrome is at risk of developing more somatic complications and psychic or mental disorders. However, their families are better documented and present a better social and health experience vis-à-vis this handicap. In this phase, multidisciplinary collaboration in the care setting is essential, as in all previous phases, to prevent and minimize probable health problems and above all to facilitate progress in the care pathway.

Aging in people with Down syndrome meets the same criteria as in the general population. It is multi-factorial and variable depending on the individual, modulated by the innate, the state of health and the environment.

The behavioral phenotypes and cognitive domains to be evaluated and explored in people with Down syndrome are multiple and adapted according to the different phases of the subject's life. The most commonly used psychological tools in practice are:

- The Wechsler intelligence scale for children, adolescents, and adults (WPPSI-IV, WISC-V, WAIS-IV),
- The psycho-educational profile (PEP III, AAPEP),
- The attentional functions (KITAP, TeaCh),
- The Executive functions (BRIEF, subtests de la NEPSY-II).

This type of evaluation must be continuous to explore the evolution of language, cognition, and behavior in order to adapt psychological and educational care to the subject with Down syndrome. Their parents must be informed of the results of these assessments in order to be able to assess the progress and benefits of the management procedures undertaken.

4- The Psychiatric Care of Trisomy 21 Inventory

The psychiatric care of the subject with the trisomy 21 disability responds to a well-defined protocol which provides information on the elements of prevention and psychiatric follow-up in the event of the appearance of mental disorders. The mental disorders that a Down syndrome person can have during his life are numerous and not necessarily present, we quote: sleep disorders, cognitive and behavioral disorders, anxiety disorders, mood disorders, psychotic disorders and rarely addictive disorders. The cognitive disorders are predominant in trisomy 21 and concern short-term and long-term verbal memory as well as attentional functions. Learning abilities and implicit memory are rather strong points in people with Down syndrome. However, it should be noted that there are very few studies on the prevalence of mental disorders in people with Down syndrome.

The presence of associated psychic disorders is difficult to observe or quantify before the age of three. It is therefore important not to rush a psychiatric diagnosis because of late acquisitions in the cognitive development of the subject with Down syndrome. It is therefore recommended to stimulate young children with Down syndrome as early as possible by integrating them into child care centers for early socialization.

We have observed in our practice as psychiatrist, subjects with Down syndrome are rarely brought back to psychiatric consultations for the management of their psychic disorders comorbid with their handicap. This is due to several factors including:

- Some signs of disability mask some symptoms found in mental disorders;
- Symptoms of mental disorders may be atypical and difficult to diagnose due to disability;
- The family circle and the therapists neglect the clinical signs of psychic suffering to the detriment of the handicap;
- The person with trisomy 21 disability finds it difficult to verbalize their suffering, whether psychic or somatic. Indeed, it is described that people with Down syndrome find it difficult to describe their pain, whether physical or psychological. Although people with Down syndrome have a pain sensitivity threshold described as higher than in “ordinary” people. In some cases, they don't report the pain, or they don't specify where it hurts. This is a false numbness that reflects difficulties in expressing pain more than decreased sensitivity or nerve conduction. This phenomenon is increased by facial hypotonia which can mask the expressions and mimicry of Down syndrome.

To this end, it is important that the medical training of psychiatric practitioners integrates in detail the particularities of the various psychiatric disorders that a person with

Down syndrome may have during his or her life, in order to diagnose them in time and take care of them.

5- Conclusion

Lionel Penrose says “a society should be judged on how it cares for its less favored members” (Rondal, 2010). Among these members are people with disabilities, in particular people with Down syndrome. The management of this type of disability requires human resources, including both the involvement of people close to people with trisomy 21 and the skills of professional health and social specialists. It also engages material and temporal means for the success of the early detection of the syndrome and its probable complications which affect the soma, the psyche, and the socius of the trisomic.

It has been proven that improving the quality of life of people with disabilities, including trisomy 21, depends primarily on the primacy of screening and the quality of prevention and management of secondary pathologies. It is a question of countering the processes of unsuitable care which can cause double health and social suffering, affecting the subject with Down syndrome and his close entourage.

References and Referrals

Chapman, R., & Hesketh, L. (2000). Behavioral phenotype of individuals with Down syndrome. Mental Retardation and Developmental Disabilities Research Reviews, 6, 84-95.

Comblain, A., & Thibaut, J.-P. (2009). Approche neuropsychologique du syndrome de Down. In Traité de neuropsychologie de l’enfant (pp. 491-524).

Curry, C. J., Stevenson, R. E., Aughton, D., Byrne, J., Carey, J. C., Cassidy, S., Opitz, J. (1997, November 12). Evaluation of mental retardation: Recommendations of a consensus conference. American Journal of Medical Genetics, 72(4), 468-477.

Dalla Piazza, S., & Dan, B. (2001). 21. Le syndrome de Down. Handicaps et déficiences de l'enfant, 317-329.

Dolk, H., De Wals, P., Gillerot, Y., Lechat, M., Aymé, S., Beckes, R., Stoll, C. (1990). The Prevalence at birth of Down syndrome in 19 regions of Europe 1980-86. Key issues in mental retardation research, 3-11.

Down, J. L. (1866). Observations on an ethnic classification of idiots. Lecture Reports (3), 259-262. London Hospital Clinical.

Ecalle, J., & Magnan, A. (2002). L'apprentissage de la lecture : Fonctionnement et developpement cognitifs. Paris: Armand Colin.

Futura-Santé. (2021). Définition/ Trisomie 21-Syndrome de Down/ Futura Santé. Retrieved Fevrier 27, 2021, from Futura Santé: https://www.futura-sciences.com/sante/definitions/medecine-trisomie-21-7719/

Larouse médical. (2002). Larousse.

Moudjahid, J. E. (2010). Près de 2 millions de personnes en Algérie vivent avec un handicap. Algérie.

Rondal, J.-A. (2010). La Trisomie 21 perspective historique sur son diagnostic et sa comprhénsion (Mardaga ed.). Belgique.

Sanlaville, D., Touraine, R., & De Fréminville, B. (2020). Protocole National de Diagnostic et de Soins (PNDS) Trisomie 21. CHU de Lyon- Centre de Référence CLAD Sud-Est ; CHU de Saint-Etienne - Centre de Compétence CLAD Sud-Est, Région Sud-Est.

Vaessen, S., Daron, A., Dubru, J., Ebetiuc, I., Leroy, P., & Misson, J. (2012). Aspects neurologiques associés au Syndrome de Down. Journal du Pédiatre Belge, 14(2), 40-43.

Verloes, A. (2015). Trisomie 21: du phénotype au génotype. INSERM.