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Clinical Endocrinology and Metabolism

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Case Report

The Diagnosis Wandering of the Williams-Beuren Syndrome: A Case Report

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Received date: March 01, 2023; Accepted date: March 13, 2023; Published date: March 22, 2023

Citation: Bouchra Aabbassi, Fatiha Manoudi, (2023), The Diagnosis Wandering of the Williams-Beuren Syndrome: a Case Report, *Clinical Endocrinology and Metabolism*, 2(2) **DOI:**10.31579/2834-8761/13

Abstract

Williams or Williams-Beuren syndrome (SWB) is a rare, sporadic and non-hereditary genetic disease, relating to a chromosomal microdeletion at 7q11.23. It is a disorder that affects multiple systems. Many children with this disease, li all rare diseases, do not benefit from early diagnosis. This leads to a real diagnostic wandering and a rather disjointed ca circuit. We deploy a case of a girl in whom the diagnosis of SWB syndrome was made by the child psychiatrist at the ag of six despite having consulted the front line many times long before.

Keywords: l-arginine, omega-3 polyunsaturated fatty acids, cerebral ischemia

Introduction

Williams or Williams-Beuren syndrome (SWB) is a rare, sporadic and non-hereditary genetic disease, relating to a chromosomal microdeletion at 7q11.23. It is a disorder that affects multiple systems. It is essentially characterized by facial dysmorphism, cardiovascular malformations and a specific neuropsychological profile. However, it remains unknown to clinicians. Many children with this disease, like all rare diseases, do not benefit from early diagnosis. This leads to a real diagnostic wandering and a rather disjointed care circuit. This will not be without consequences for the child. We deploy a case of a girl in whom the diagnosis of SWB syndrome was made by the child psychiatrist at the age of 06 despite having consulted front-line clinicians many times long before.

Case Report : Aya is six years old child who consults in child psychiatry, accompanied by her two parents for school learning difficulties. She is still in the middle section of kindergarten. Parents who have consulted several pediatricians since the age of three for global psychomotor retardation. The little girl benefited from rehabilitation sessions in psychomotricity and speech therapy but with “timid progress”, says the father. Apart from these complaints, Aya had no personal or family history of organic or psychiatric illness. Nevertheless, a deafness assessment was carried out three years ago to explore the origin of the oral language delay. The current psychiatric evaluation finds a jovial girl, with familiar contact, with an endearing gaze, her language is still poorly articulated. She is motorally unstable but without noticeable clumsiness. Regarding learning, Aya does not yet have access to reading or writing. His drawing is reduced to scribbles. Furthermore, we find no signs of depression, anxiety or environmental dysfunction. The parents seem committed and invested in their little girl to the point of postponing any decision to have a second child for fear of abandoning Aya! The girl has a face with a small chin, a wide and full-lipped smile, a wide forehead and a flat nasal bridge. These dysmorphic and behavioral traits are reminiscent of the Williams-Beuren syndrome. Genetic counseling has been requested. He confirms William-Beuren's diagnosis. Cardiovascular, endocrine, neurological and neurosensory (auditory and visual) somatic evaluation revealed slight weight loss and supravalvular aortic stenosis without functional repercussions. The neurocognitive assessment concluded to a mild to moderate intelectuel disability. In light of these assessments, multidisciplinary care was proposed for Aya: regular pediatric follow-up combined with developmental child psychiatry follow-up with parental guidance, stimulation and awakening psychotherapy, speech therapy sessions focused on expressive language and writing, psychomotricity sessions with school facilities. Aya was able to join an association for children carrying Williams Beuren. She is currently in the first year of elementary school. Apart from difficulties in managing her emotional outbursts, she progresses in school.

Discussion: SWB is a developmental anomaly described for the first time in 1961, whose genetic cause was identified in 1993: a microdeletion of the chromosomal region 7q11.23 leading to the loss of an allele of 26 to 28 genes on one of the two chromosomes 7 [1, 2]. One of these genes is of elastin and other genes involved in hypercalcemia, growth retardation, intellectual disability and behavioral disorders. Most patients have the same deletion span, but few individuals have smaller or larger deletion fragments, which would explain the great heterogeneity of the clinical pictures and the non-specificity of the facial dysmorphism which can induce a diagnostic delay. It has been estimated that the prevalence of WBS is approximately 1/75001/20,000 [2,3]. Clinical manifestations include:

• Cardiovascular disease: elastin defect arteriopathy, supravalvular aortic stenosis, stenosis of the branches of the pulmonary artery, arterial hypertension.

• An evocative facial morphology: a broad forehead, bitemporal narrowing, periorbital fullness, an iris with star and/or lace patterns, a short, upturned nose with a bulbous tip, a long philtrum, a wide mouth, full lips and mild micrognathia. This dysmorphism is inconstant; wich can explain the deviations in the clinical assessment.

• Retarded growth of wight et height

• Neonatal hypercalcemia in 15% of cases

• Digestive disorders such as gastroesophageal reflux in early childhood or colonic diverticulosis at any age

• Moderate intellectual disability

• A specific cognitive profile characterized by great difficulties in the visual-spatial domain contrasting with an apparently correct language

• A rather specific behavior, of the hypersociable type, approaching others easily. Children have a hypersensitivity to noise and a disposition for music, great language faculties, as well as a good auditory memory.

• Anxiety remains the most common psychiatric comorbidity in 54% of patients. Psychotic symptoms are exceptional but may occur in adolescence.

The circumstances of discovery in childhood are generally on the occasion of a:

• Heart disease, often revealed neonatally in the presence of a murmur or signs of cardiac decompensations.

• Neonatal hypercalcemia

• Psychomotor retardation

It is the association with the morphology of the face, and often with growth retardation and/or eating difficulties that orient the diagnosis. When the diagnosis is not made in early childhood, the characteristic behavioral profile can oriente the clinicians to diagnosis the syndrome, towards the 5-6 years old. The positive diagnosis is, since 1991, confirmed by chromosomal analysis called FISH test [4]. The standard caryotype is often normal. The diagnosis of the disease is the responsibility of the medical specialist (pediatrician, cardiologist geneticist). But the initial assessment and the overall care of the patient are based on multidisciplinary cooperation, and involve doctors from several disciplines: general pediatrician, cardio pediatrician, neuropediatric Ian, clinical geneticist, cardiologist, nephrologist, ophthalmologist, endocrinologist, orthopedist, psychiatrist, hepato-gastroenterologist, ENT doctor, radiologist, biologist, rheumatologist, rehabilitation doctor. And other professionals like nurse, physiotherapist, psychologist, psychometrician, speech therapist, dietitian, educator, dentist, orthodontist, orthoptist, social worker, educator. The announcement of the diagnosis is an essential step in the care process. It must be the subject of a dedicated consultation and includes: the explanation of the diagnosis, the planning of the care and follow-up, with the description of the multidisciplinary team that will provide it. Information on the natural history and prognosis of the SWB syndrome, on the treatments prescribed, their possible adverse effects; on the need for regular follow-up and on the planning of the examinations necessary for diagnosis, monitoring of the disease or screening for possible complications. It is also essential to emphasize that the parents are not the cause of this trouble; it is an accidental disease that can occur in any family. This information reassures the parents and frees them from the unconscious pression of guilt towards their child, as was the case with our patient. Aya's parents were more engaged and collaborative in caring for their child after the announcement. They even thought about having a new child in the same year of follow-up, while the couple was reluctant to this idea before. However genetic counseling is essential. The risk of recurrence is very low, if it is verified that the genetic anomaly appeared "de novo" in the patient [3]. The risk of germinal mosaic (presence of several mutated gametes in one of the parents). For a new pregnancy, prenatal diagnosis by FISH analysis of the SWB critical region on fetal cells (carried out from a chorionic villus biopsy or amniocentesis depending on the term of the pregnancy) may be proposed. The parents of our patient were sent to genetic counseling to accompany the decision to procreate. Currently, there is no cure for Williams syndrome. In childhood, the essential is to treat cardiac defects and hypertension by pediatric cardiology teams aware of this pathology, of hypercalcemia by suppressing the supply of vitamin D, a diet low in calcium, and /or injectable pamidromete.The management of nephrocalcinosis, or hypercalcemia and/or hypercalciuria requires specialized opinion from nephropediatrics. ophthalmological follow-up is necessary. The educational care of children must be done in a multidisciplinary framework involving pediatricians, psychometrician, speech therapist and psychologist. A neuro-psychological and behavioral assessment is essential to guide the individual management of learning disabilities, as well as attention and anxiety disorders which are frequent. Each stage of development or each difficult phase requires support. It is an ongoing process. The support of child and parents by the multidisciplinary team is essential, and the patient association can be extremely useful. A child who is well taken care of and accompanied on the various learning paths, supported by the educational and medical teams as well as by the family, has every chance of becoming an adult assuming an independent life. Individual and family psychotherapy, whether supportive or more structured, finds its place in the child's therapeutic arsenal. Psychotropic drugs are often necessary for behavioral disorders, especially in cases of attention deficit or anxiety disorders. Relaxation techniques can also help with anxiety [3,4]. On the developmental level, somatic and psychiatric follow-up must be programmed at an annual rate until adulthood. In particular neuropsychological and/or psychiatric evaluations looking for behavioral disorders, environmental and school adaptation difficulties, anxious or depressive comorbidity and more rarely, but not exceptionally, psychotic derivation as recently described on literature in entering adolescence [5,6]. . The prognosis of this disease is linked to the degree of organic damage and psychiatric comorbidities, hence the need for early diagnosis and treatment. Fortunately for our patient, the medical case was manageable despite the diagnosis delay.

Conclusion:

Rare diseases continue to elude first-line care circuits for early screening and diagnosis. Much remains to be considered to overcome this difficulty, in terms of raising awareness and training professionals, setting up evaluation strategies and above all the establishment of care circuits for these young patients and their parents who find themselves often without response to requests taken lightly by the medical staff. When in truth, these are serious diseases in terms of development and prognosis.

Competing interests: the author declares no competing interest.

Acknowledgements: no acknowledgements.

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