Annals of Case Reports & Reviews

Case Report

doi: 10.39127/2574-5747/ACRR:1000195

Aabbassi B and Benali A, et al. Annal Cas Rep Rev: ACRR-195

A Case of Angelman Syndrome in Child Psychiatric Consultation

Bouchra Aabbassi*, Abdeslam Benali

Research team of mental health, University Hospital Centre of Mohamed VI, Marrakesh, Morocco

***Corresponding author:** Bouchra Aabbassi, child psychiatric, University Hospital Centre of Mohamed VI, Marrkesh, Morocco. Email : aabbasi-iam@hotmail.com

Citation: Aabbassi B and Benali A (2021) A Case of Angelman Syndrome in Child Psychiatric Consultation. Annal Cas Rep Rev: ACRR-195.

Received Date: 21 January 2021; **Accepted Date:** 25 January 2021; **Published Date:** 31 January 2021

Abstract

"Angelman syndrome" also known as "Happy Puppet Syndrome", is a rare genetic disorder linked to the lack of function or contribution of UBE3A gene located on the maternally derived allele of 15q chromosome. This disorder is characterised by severe psychomotor delay and severe intellectual disability, with absence of language, jerky movements, frequent laughter or smiling and almost constantly epilepsy. This syndrome is causing severe motor and intellectual disabilities. It affects deeply the life of the child and his family. We report the case of a 2.6 years-old-girl brought, first, in child psychiatry department and who is being diagnosed as "Angelman Syndrome".

Keywords: *Angelman syndrome, psychomotor disabilities, delayed developpemental.*

Background

Angelman Syndrome "," Hilarious puppet syndrome "," Happy puppet syndrome "or" Joyful puppet "are all names for a rare neurogenetic disease. This is a genetic defect linked to the lack of function or contribution of the UBE3A gene located on maternal chromosome 15 [1, 2]. Clinically, this anomaly is manifested by significant psychomotor retardation and severe intellectual disability, with absence of language, easy fits of laughter and almost constant epilepsy [1-3]. This syndrome is at the origin of a severe handicap, both motor and intellectual. It has a profound impact on the life of the child and that of his family.

Case presentation

L. is 2 years. She is the only daughter of a married couple. We receive her in child psychiatric consultation for a delay in psychomotor acquisitions. The clinical and ultrasound monitoring of the pregnancy did not conclude to any abnormalities. At birth, L. weighed about 2.5 kg with a normal size.At the age of 6 months, she does not follow her gaze and responds little to the stimuli of those around her, she is easily irritable and has sleep problems. Yet she is "jovial" and smiles "very often". Growing up, L. did not acquire grating, walking or speech. At the age of one year, the girl begins to have myoclonic crises for which she receives occasional prescriptions for Soduim Valproate without regular pediatric follow-up.

The developmental disability becomes noticeable by 2 years of age. The parents decide to consult child psychiatrist . L, with an enchanting gaze and a catchy smile, is constantly held in her mother's arms. She remains silent. L. depends entirely on the care of his mother.

We referred L. to pediatric neurologist consultation. The doctor performs the following assessment: Brain MRI (normal), EEG (background activity replaced by slow waves at 2-3 Hz synchronous and symmetrical triphasic), karyotype (46 XX without detectable abnormalities), Molecular cytogenetics (deletion at the Angelman locus in 15q11. 2 with the UBE3A probe). Visual evoked potentials show decreased activity of the visual pathways with conduction disturbance in both eyes.

The pediatrician retained the diagnosis of Angelman's syndrome and adjusted the doses of Soduim Valproate. We set up psychomotor rehabilitation, speech therapy and parental guidance. We also recommended socialization in kindergarten with educational support for the next school year. L. has been doing well ever since. Although she doesn't say a word, she is understood when she signs or expresses herself with the body.

The parent's great fear remains L's future, will she grow up well? Will she one day be autonomous? What type of school will be able to accommodate What educational and professional future can we predict?.

Discussion

In 1965, Harry Angelman, an English pediatrician, reported the clinical observation of three children and defined "Puppet Children" or "Puppet Children". In 1982, this name was changed to "Angelman Syndrome".

It is a neurogenetic disorder that affects the 15q11-q13 region of the long arm of maternal chromosome 15 (deletion, absence, point mutation, non-expression of the UBE3A gene) [1-4]. Its global prevalence is estimated between 1 / 20,000 and 1 / 10,000. Children of both sexes can be affected [1-4]. The diagnosis is clinical. The history of the disease begins around 1 year of age. These children almost constantly present with severe intellectual disability with slow and delayed psychomotor development and language absent or reduced to a few words (comprehension is however better than expression) [4-7]. Balance disturbances with unsteady gait, hyperactivity, hyperexcitability and reduced attention with very easy smiles and laughter have also been reported as constant signs [4, 5]. Epileptic seizures usually begin before 3 years of age, with particular electroencephalogram abnormalities characterized by slow waves of great amplitude and bursts of wave peaks producing an aspect of predominantly frontal delta triphasic activity [1, 3]. Myoclonus of the limbs and extremities giving the impression of trembling and blinking of the eyes and very severe sleep disturbances have been reported. There are somme particular features of the face, with a big smile, wide teeth and a flat occiput due to hypotonia and therefore to the delay in acquiring the sitting position, hypo pigmentation of the skin, hair, strabismus, excessive drooling and a tendency to put everything in the mouth [2, 6].

Also,the "angelic" behavior of these children is very characteristic: they are very social and seek contact with adults, but their attention span is very limited. They particularly enjoy water, music, photos, and musical or light toys [3].

In adolescence, one should fear the onset of scoliosis and deformities of the joints in the feet and knees. Sometimes obesity with a tendency to bulimia [3, 7].

Genetic analyzes confirm the diagnosis in around 90% of cases (The karyotype associated with the so-called FISH technique) [1, 2, 6]. When no deletion is detected, further genetic analysis is performed to determine which markers are present on the genes in the 15q11-q13 region. In one in ten cases, no genetic abnormality can be detected [3]. A fundus examination is often done . Visual evoked potentials (VPE) measurement can be used [1, 2].

There is currently no treatment for Angelman syndrome [1-4]. The care is aimed at the development of the child, his stimulation and support for those around him. Up to 3 years of age, socialization is favorable and allows contact with the ordinary environment. After 3 years, it is also possible to obtain kindergarten education, in the presence of a school assistant. Medical treatments can improve the various manifestations, in particular epilepsy, behavioral and sleep problems [3].

In all cases, psychological guidance seems essential to support parents. Parent groups and associations seem fundamental to share experiences.

Complete autonomy is never reached [1, 3, 4]. In adolescence, most of the difficulties persist, but there does not appear to be any neurological worsening. Sexual development is delayed. Regular medical monitoring should be put in place to monitor the appearance of certain clinical signs: skeletal deformities, weight gain, ophthalmological problems and thyroid dysfunction. Later, young adults continue to progress, they sleep better, and seizures more often decrease.

If the parents of a child with Angelman syndrome are planning a new pregnancy, genetic counseling is recommended because of the risk of recurrence in 0 to 50% depending on the genetic defect involved [1, 6].

Conclusion

Angelman syndrome is a rare and severely disabling disease. Beyond the psychoeducation and the various re-educations such as physiotherapy, speech therapy and psychomotor skills that we offer, the care is based on information and family support.

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