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Aicardi-Goutières Syndrome: About a 7-Month-Old Infant

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

The Aicardi-Goutières syndrome is an encephalopathy characterized by an arrest of psychomotor development in the first months of life, spastic tetraplegia spastic tetraplegia, abnormal movements and acquired microcephaly. Neuroimaging reveals calcifications of the basal ganglia and cerebrospinal fluid (CSF) study shows lymphocytosis. Our patient is a 7 month old child, from a 1st degree consanguineous marriage. She presents a psychomotor delay with hypotonia, nystagmus and convulsive myoclonic seizures. Cerebral CT shows an active tri ventricular hydrocephalus with multiple parenchymal calcifications with multiple cerebral parenchymal calcifications and basal ganglia of infectious appearance. The intercritical sleep EEG was unremarkable. Ophthalmological examination, including an EPI, revealed a probable retinal dystrophy predominantly on the scopic system. Brain MRI shows a mesencephalic atrophy of the annular protuberance and increased volume of the volume of the cisterns of the posterior cerebral fossa. The dosage of interferon alpha in the CSF is increased to 113 pg /ml.

Keywords: Aicardi-Goutières syndrome; psychomotor development; encephalopathy.

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1. INTRODUCTION

Aicardi-Goutières syndrome is a progressive encephalopathy characterized by arrested psychomotor development in the first months of life, spastic tetraplegia, abnormal movements, and acquired microcephaly. Neuroimaging shows calcifications of the basal ganglia and cerebrospinal fluid (CSF) study shows aelymphocytosis.

2. OBSERVATION

Our patient is a 7 month old child, from a consanguineous marriage of the 1st degree and a 29 years old mother, G1P1, grouping B+, pregnancy well followed, the delivery took place by caesarean section for exceeding the term, birth weight= 2kg600 with immediate cry, put under exclusive breastfeeding. She presented a psychomotor delay with hypotonia, nystagmus and myoclonic convulsive seizures. Cerebral CT shows an active tri ventricular hydrocephalus with multiple cerebral parenchymal calcifications and basal ganglia of infectious appearance.

The intercritical sleep EEG was unremarkable. Ophthalmological examination including an EPI revealed a probable retinal dystrophy predominating on the scopic system. Brain MRI shows a mesencephalic atrophy with atrophy of the annular protuberance and increased volume of the cisterns of the posterior cerebral fossa. The interferon alpha level in the CSF is increased to 113 pg /ml. The genetic study is in progress.

On these clinical, neuroradiological, ophthalmological signs and biochemical results, we concluded to an Aicardi-Goutières Syndrome.

3. DISCUSSION

Aicardi-Goutières syndrome (AGS) was first described in 1984 by Jean Aicardi and Françoise Goutières two eminent pediatric neurologists working at Necker Hospital in Paris, France [1]. They reported eight children from five families with an early onset of central nervous system disorder with bilateral spasticity and dystonia and acquired microcephaly. The course was rapid to death.

It affects mainly female males, with a few exceptions, men with Klinefelter syndrome (47, XXY) because there is a hypothesis that this disease is the result of a de novo mutation on the X chromosome [2].

This syndrome was defined as the triad of infantile spasms, agenesis of the corpus callosum and chorioretinal lacunae [3]. The syndrome is genetically heterogeneous with mutations in 7 genes, including TREX1, RNASEH2A, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1 and IFIH1 (interferon-induced helicase c domain-containing protein 1) [4].

Aicardi-Goutières syndrome (AGS) is an inflammatory mendelian leukoencephalopathy Aicardi-Goutières syndrome (AGS) is an inflammatory mendelian leukoencephalopathy, part of the type I interferonopathies - a group of neuroinflammatory phenotypes associated with increased type I interferon (IFN) signaling, characterized by basal ganglia calcification, white matter abnormalities, and cerebrospinal fluid (CSF) lymphocytosis. It is a rare autosomal recessive neuro-pediatric syndrome with an encephalopathy beginning before the age of one year and of progressive aggravation, leading to a loss of acquisitions then a severe intellectual and psycho-motor deterioration.

The symptoms evolve over a few months with the appearance of microcephaly and a pyramidal syndrome before stabilizing. However, more moderate forms have been described with onset after 1 year of age, relative preservation of language and cognitive functions, and normal head circumference. Inter- and intra-familial phenotypic variability is usually observed.

Most of patients develop normally until 3 months of age, and then, epileptic seizures and developmental delay start to emerge [5], in our case the baby developed spasm-like epileptic seizures at the age of seven months.Drug resistant epilepsy is usually seen in severe cases, along with severe cognitive impairment and reduced life expectancy [6].

The diagnosis of Aicardi-Goutières syndrome is still often misunderstood. Epileptic spasms are often early, as in our patient's case, and as early as the neonatal period in 18% of cases [7]. On EEG, true hypsarrhythmia remains rare, the most typical aspect being asynchronous pseudoperiodic discharges on both hemispheres [8]. Chorioretinal lacunae are found and are yellowish white, rounded with a clear

unpigmented border. They are most often multiple and bilateral and are pathognomonic of the syndrome. Other ocular abnormalities may be associated with chorioretinal lacunae such as unilateral colobomas of the optic nerve, iris and peripapillary choroid [9], microphthalmia often unilateral, microcornea, calcified cataracts, iris synechiae and pupillary membrane remnants, optic nerve hypoplasia, optic atrophy, retinal detachment, or sclerectasia [10].

CT scan shows calcifications of the basal ganglia and MRI shows suggestive hypodensities in the white matter. The increase of interferon alpha in the CSF is almost constant. Cerebral histological specimens may show microinfarcts and calcifying vasculitis of the brain and systemic vessels.

TORCH complex embryo-fetopathies, neonatal enteroviruses, Cockayne syndrome, hereditary progressive CNS degeneration with generalized intracranial calcifications are the main differential diagnoses to be ruled out [11].

The prognosis of these patients is guarded; it's varies with the severity of brain abnor- malities and underlying symptoms, and life expectancy could be severely limited from several months to only a few year.The evolution is usually towards profound mental retardation, blindness and the absence of any motor or language acquisition [12].

The treatment remains symptomatic, consisting in the management of eating disorders, psychomotor deficits, and possible epilepsy. The evolution towards a profound deterioration and death is generally severe and rapid.

4. CONCLUSION

Aicardi-Goutières syndrome is a rare encephalopathy. This observation illustrates the interest of making the diagnosis and the importance of genetic counseling because there is a risk of recurrence in a subsequent pregnancy. Prenatal diagnosis can be made by molecular biology on amniotic fluid or trophoblast.

CONSENT

As per international standards, informed and written parental consent was collected and preserved by the authors.

ETHICAL APPROVAL

As per international standards, written ethical permission was collected and preserved by the author.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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