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Original Research Paper

Medicine

CORPUS CALLOSUM AGENESIS - CLINICAL CASE PRESENTATION

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ABSTRACT

Objectives: Describe the causes, clinic, diagnosis, treatment of this disease, as well as its clinical follow-up.

Method: A retrospective study was carried out, a clinical case from detection of malformation in the prenatal stage taking place at birth, up to its postnatal clinical follow-up.

Results: Female gender patient with prenatal diagnosis at 25 weeks and postnatal by imaging exams (US, CT, MRI, spectroscopy) of Corpus Callosum associated with subclinical epileptiform discharges, hypothyroidism and strabismus who is maintained in clinical pharmacological treatment + Physiotherapy evidencing a noticeable improvement in the control of upper limb movements and relationship with their social environment at one year of age.

Conclusion: Uncommon congenital malformation, its presentation can be isolated or associated with other malformations. With variable clinical presentation. The diagnosis can be prenatal or postnatal using imaging techniques, with MRI being the one with the highest sensitivity and specificity.

There is no cure or standardized treatment, sometimes the intervention of some medical specialties is required for its symptomatic treatment.

KEYWORDS : Agenesis, corpus callosum, malformation.

INTRODUCTION

Corpus callosum agenesis (CCA) is a congenital defect, characterized by partial or total absence of the corpus callosum. It can occur as an isolated defect or in combination with other brain abnormalities, including Arnold-Chiari malformation, Dandy-Walker syndrome, schizencephaly (grooves or indentations in brain tissue), and holoprosencephaly (lack of division forebrain into lobes).

In women it can have a sex-specific disease called Aicardi syndrome that occurs with a triad: agenesis of the corpus callosum (total or partial), typical chorioretinal gaps and infantile spasms

The signs and symptoms of the disorder range from subtle or mild to severe, depending on the associated abnormalities. Intelligence may be normal with a slight compromise of skills that require relating visual patterns or, in the case of children with more severe malformations, there may be intellectual retardation, seizures, hydrocephalus and spasticity.

The prognosis depends on the degree and severity of the malformations. ACC does not cause death in most children. Mental retardation does not worsen. Although many children with the disorder have average intelligence and lead normal lives.

There is currently no standard cure or treatment for ACC. Treatment is done to manage the symptoms the person has.

METODOLOGIA

A retrospective study was carried out, a clinical case from detection of malformation in the prenatal stage taking place at birth, to its postnatal clinical follow-up.

The information and images obtained belong to the medical personnel in charge of the case whose reinforcements rest in the statistical package Excel, Word and JPG.

PRESENTACION DE CASO CLINICO:

28-year-old pregnant woman, with no known pathological history. Who in a control at week 25 of gestation is evidenced by ultrasound Fetal Bilateral Ventriculomegaly

At 38 weeks of gestation, a cesarean delivery was performed due to pelvic cephalous disproportion (DCP), newborn female gender, APGAR: 8-9, height: 50cm, head circumference: 36cm, chest circumference: 35cm, presence of primitive reflexes, with apparent hyperreflexia.

She was admitted to the Neonatal Intensive Care Unit at 72 hours of life for phototherapy (hyperbilirubinemia> 17 mg) where she remained for 2 days.

Brain Simple Computed Axial Tomography (CT) performed at 10 days of age, dilatation of the temporal and occipital horns of the lateral ventricles was evident, with little convergence between them (data suggestive of corpus callosum agenesis).

He received an evaluation by a Neuropediatrician who, upon physical examination, found slight involuntary movements of the upper limbs, for which tests were requested:

Simple Simple Brain MRI: Signs of corpus callosum agenesis. Prominence of ventricular bodies in their posterior aspects that suggest colpocephaly. Data of delay in the myelination process.

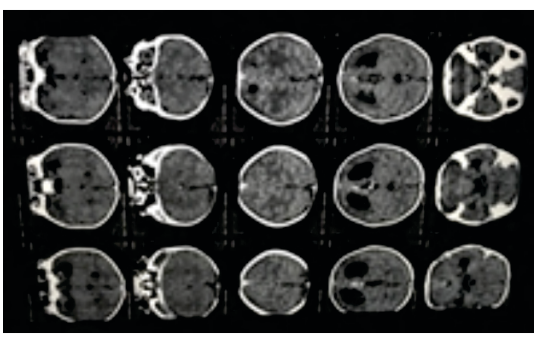
- Spectroscopy: Suggested alteration or immaturity in myelin precursors and decreased neuro-axonal population.
- Electroencephalogram (EEG): Focused discharges of left frontal distribution.
- Thyroid Ultrasonography: normal with T3: 13

With these results, treatment with Valproic Acid 75 mg orally every 12 hours + levothyroxine 17 mcg orally 3 times per week + early stimulation and rehabilitation was started.

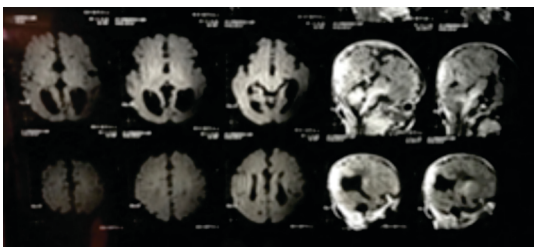
A new control was performed at 8 months of age with EEG in wakefulness and induced sleep, showing occasional generalized paroxysms of slow waves in the high-voltage polymorphic delta range.

In addition, due to the presence of strabismus, an ophthalmologist was evaluated, who found no associated pathology and indicated treatment with eye patches for 3 months.

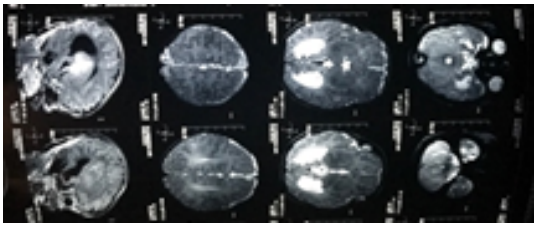
At one year of age after the mentioned pharmacological treatment (with dose correction of Ac. Valproico: 125 mg PO w / 12h and levothyroxine: 31.5 mcg / day) associated with physiotherapy sessions (early stimulation once a week, language therapy Once a week) and occasional matronation there is marked improvement in upper limb control, allowing you to firmly grasp nearby objects, explore people's faces. The relationship with their social environment has improved, the vaccination schedule is complete for their age.



Graph1: TAC Skull



Graph2: RNM Craneo



Graph3: Spectroscopy

DISCUSSION

Corpus Callosum Agenesis is rare, estimated to occur in approximately 1.8 out of 10,000 live births. It is believed to be caused by an attack between 5 and 16 weeks of gestation (time in which the formation and development of the corpus callosum occurs) and is usually diagnosed in utero at 20 weeks of gestation.

Some authors report a higher incidence in males when ACC is isolated, (49%) of cases.

Its causes are unknown, some associate it with viral and non-viral infections (influenza, rubella, toxoplasmosis), exogenous toxic factors (cocaine, alcohol, valproate), endogenous toxic factors (lactic acidosis, hyperglycemia) and lately some researchers have associated vascular factors. Being an inherited disorder, its pattern is mostly autosomal recessive.

Prenatal diagnosis can be made by ultrasound or magnetic resonance from the 20th week of gestation, postnatal diagnosis can be carried out by performing ultrasound, computed tomography or brain magnetic resonance imaging. It is ideal that in any diagnostic scenario, prenatal or postnatal, resonance is used since it has greater sensitivity and specificity, mainly to find associated anatomic abnormalities.

Each patient is a unique case since the signs and symptoms are very varied depending on the type of defect and associated problems. Some have no apparent problems; however, behavioral similarities have been observed that are especially noticeable from adolescence.

- Delayed motor and language development or cognitive development
- Poor motor coordination
- High sensitivity to tactile stimuli, such as food texture
- High tolerance to pain

There is no specific treatment, initially rehabilitation treatment is indicated such as: speech therapy, physical therapy, psychomotricity, occupational or educational therapy, accompanied by parent training and advice to teachers; since despite ACC the processes of neural plasticity are intact.

Discard concomitant malformations or associated genetic syndromes, which will allow a therapeutic approach, achieving a better prognosis for the patient.

Treatment should be started as soon as possible to take advantage of the plasticity of the nervous system.

CONCLUSION

Corpus Callosum Agenesis is a very rare tumor, it may be partial or complete, with isolated presentation more frequent in males or associated with other malformations (Aicardi syndrome the most frequent and with prevalence in females). Its clinical characteristics are varied, which makes each patient a unique case. The diagnosis can be prenatal or

postnatal by ultrasound, computed tomography or magnetic resonance imaging, the latter being the most sensitive and specific, with the greatest benefit in detecting associated anatomical abnormalities.

Currently there is no cure or standardized treatment for this pathology, in some cases it requires the intervention of some medical specialties to achieve control of signs and symptoms.

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