AICARDI SYNDROME: A CASE REPORT

OCAMPO-VÁZQUEZ CELIA M.D.
HIDALGO-VÁZQUEZ MÓNICA M.D.
GONZÁLEZ-MORÁN ROCCO M.D.
GARZA-MORALES SAÚL M.D.

HOSPITAL ESPAÑOL DE MÉXICO
PRESENTATION OF CASE

PERINATAL HISTORY

- Female product of a 38 week pregnancy with adequate prenatal control and no structural alterations reported in ultrasounds. She was born by an elective cesarean section receiving routine care. The birth weight was 3150 grs and APGAR scores were 9 and 9 at 1 and 5 minutes. She was discharged from the nursery as a healthy girl.

ONSET OF SYMPTOMS

- At 4 days of life, she presented seizures characterized by eye deviation, spasticity and tonic clonic movements of upper limbs with no apparent trigger factors and no familiar history of epilepsy.
- There was no history of fever, vomiting, poor feeding or trauma.
INITIAL ASSESSMENT AND STABILIZATION

- She was admitted to the ICU for diagnosis and treatment. An MRI and electroencephalogram were performed reporting agenesis and hemiatrophy of the right hemisphere, plus a disorganized and asynchronous rhythm with paroxystic events, which originated in both frontotemporal lobes.
- She was given Levetiracetam and was discharged asymptomatic.
- Two months later seizures worsened so Valproate and Clonazepam were added to the treatment, but she started presenting up to 200 crisis a day so she was readmitted for multidisciplinary management.
The patient was evaluated by the neurology, ophtalmology and genetic departments.

Due to the absence of genetic testing confirming Aicardi Syndrome the diagnosis is based on clinical features, brain MRI, ophthalmological and skeletal findings.

Results in the diagnostic tests plus epilepsy refractory to treatment highly suggested the diagnosis of Aicardi Syndrome.
Abdominal ultrasound and echocardiogram.
- Normal

Genetics
- Micro-array CGH-180K: negative
- Karyotype: 46 XX

Ophtalmologic Exam
- Right eye coloboma
- Left eye chorioretinal lacunae lesions

Vertebral X- Rays
- Thoracic hemivertebrae from t6 to t7 and butterfly vertebrae in L2 and L3.

Visual evoked potentials
- Altered conduction in the retinogeniculcortical pathway
Aicardi Syndrome is a de novo dominant X-linked neurodevelopmental disorder that affects almost exclusively females, only reporting survival in 47 XXY males. Its incidence has been estimated as 1:105,000 in the US and 1:99,000 in Europe.

- This syndrome consists of a classic triad of agenesis of the corpus callosum, distinctive chorioretinal lacunae, and infantile spasms.
- Additional manifestations such as cortical malformations, periventricular and subcortical heterotopia, cysts, coloboma, vertebral and rib abnormalities, microphthalmia, and gross cerebral hemispheric asymmetry are considered some of its supporting features.
- There is no specific genetic alteration linked to this syndrome, but there is an important association with location Xp22.
- Diagnosis is based on clinical features and image studies.