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Case Report

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# Rare Autoimmune Disease of Neonate Diagnosed with The Help of Ophthalmology

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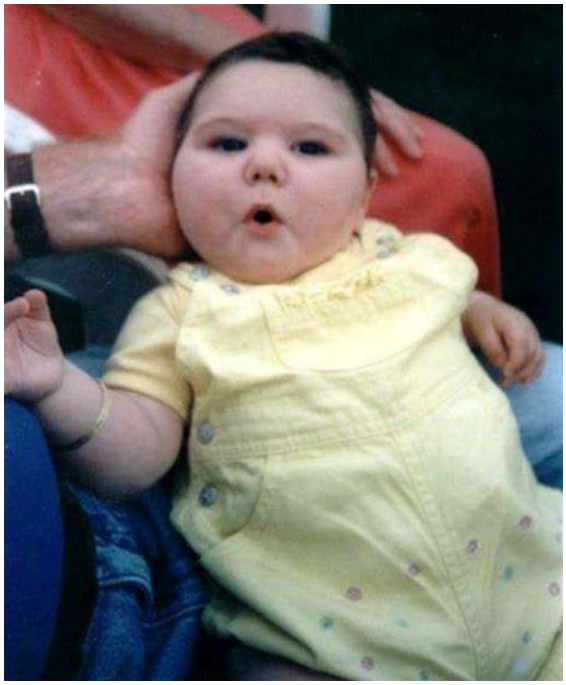
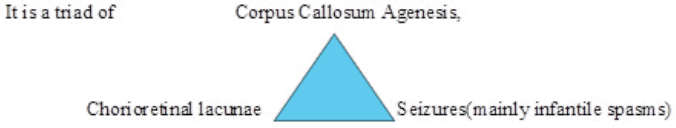


## 1. Objective

To present an interesting case of "Ophthalmic features in Aicardi Syndrome"

Aicardi syndrome is a genetic X-linked defect commonly seen in girls. The ocular finding in Aicardi Syndrome are important because they may be the initial manifestation of the disease.




## 2. Case Report

A 45 days old Female presented to the NICU with microcephaly, microphthalmos and recurrent seizures. She had a full-term Caesarean delivery. The infant also had missing 7-9 ribs, mild scoliosis and iris coloboma. There was no family history of such condition.

## 3. Discussion

Hypothetically Aicardi syndrome occurs due to spontaneous mutation at Xp22 chromosome. Though no exact etiology describes the condition. Although cases occur throughout the world; exact incidence and prevalence is unknown.

The syndrome occurs in patients of diverse clinical backgrounds and no racial predominance is noted.

Aicardi syndrome is complicated by severe mental retardation, intractable epilepsy and a resultant propensity to pulmonary complications. The condition often leads to death in the first decade. Sudden unexplained death is common. The oldest reported case was 32 years old. Average age of death in west is 8.8 years.

## 4. Clinical Features

Seizures (infantile spasms)

Global developmental delay

Microcephaly, axial hypotonia, appendicular hypertonia with spasticity.

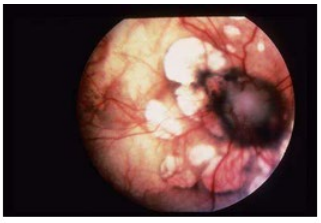
Constipation, GERD, diarrhoea, feeding difficulties.

Costovertebral anomalies such as hemivertebrae, butterfly vertebrae, rib abnormalities

Short philtrum, cleft lip, cleft palate.

## 5. Ophthalmic Features

- Microphthalmia
- Chorioretinal lacunae(hallmark)
- Retrobulbar cyst
- Cataract
- Coloboma
- Retinal detachment
- Iris synechiae
- Remnants of fetal pupillary membrane



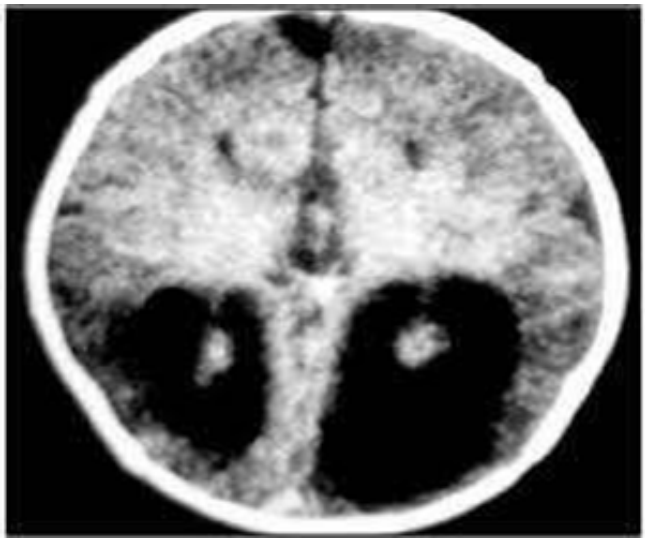
## 6. Investigations

Plain radiographs can be done for skeletal malformations.

Based on clinical suspicion, ultrasonography through anterior fontanelle is initial investigation of choice for infants.

MRI- brain with spine is gold standard investigation and is preferred. CT scan may be a helpful additional study.

EEG- can be done for typical presence of burst suppression pattern, with complete asynchrony
between two hemispheres.



## 7. Treatment

1. There is no permanent cure.
2. Seizures are treated with multiple anti-epileptic medications. Most effective treatment includes-ketogenic diet, vigabatrin, lamotrigine and topiramate.
3. Patients may benefit from specialized care with physical medicine and rehabilitation specialist as well as physical, occupational and speech therapists.

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