A Female Child with Corpus Callosum agenesis and Infantile Spasm

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A 13 week old female child presented with history of abnormal movements in the form of extension of the neck and stiffness of the body since five days of age. Brain Magnetic Resonance Imaging (MRI) and eye examination with fundus pictures are shown below:

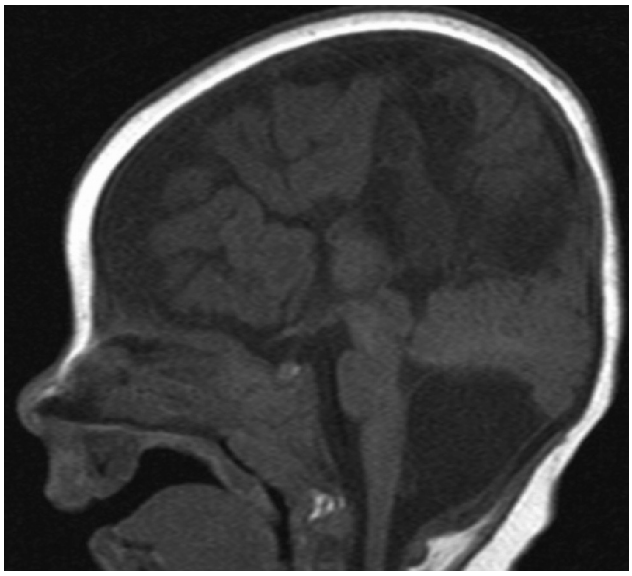

Figure 1: Sagittal T1W, midline show complete agenesis of the corpus collasum. There is posterior fossa interhemispheric cyst.

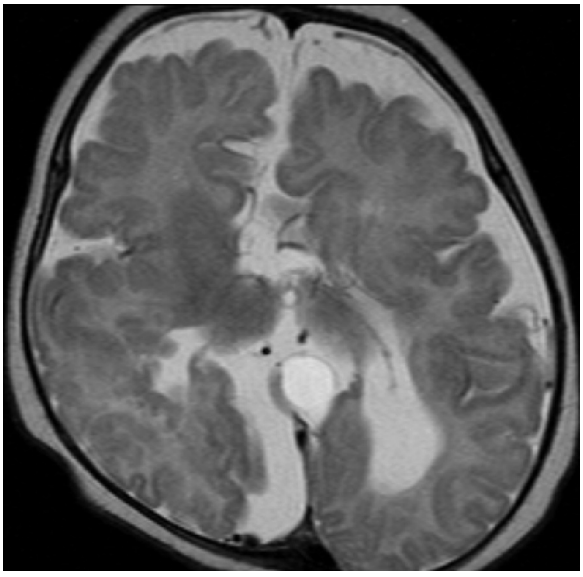

Figure 2: Corona T2W shows interhemispheric cyst, posterior fossa arachanoid cyst and periventricular heterotopic gray matter bilaterally.

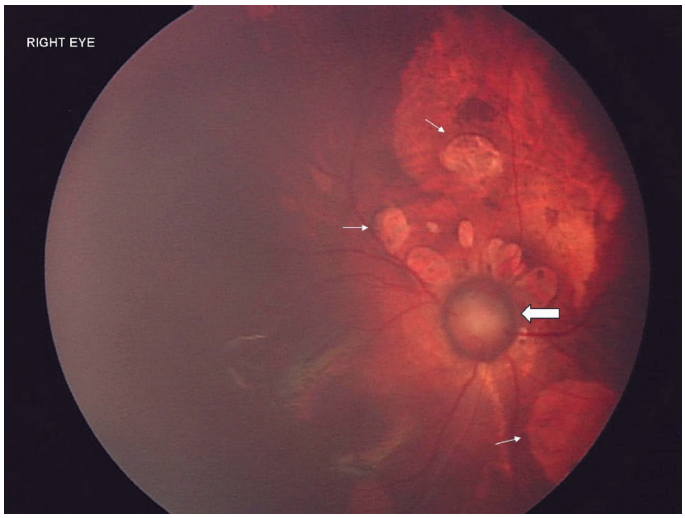

Figure 3. Small arrows show multiple well demarcated lacunae concentrated around the optic disc. Block arrow shows optic disc coloboma.

What is the most likely diagnosis?

1. Rett syndrome
2. Angelman syndrome
3. Tuberous sclerosis
4. Aicardi syndrome
5. Anderman syndrome

Answer to clinical image on page 58

The correct diagnosis of the clinical images presented above is Aicardi syndrome. The syndrome's classic triad of chorioretinal lacunae, agenesis of corpus callosum and infantile spasm is seen in this patient.

Discussion

Aicardi Syndrome is a rare x-linked dominant disorder. The disorder is phenotypically heterogeneous. [1] It presents with substantial variability in the severity of clinical features, and survival. The syndrome was first described by Aicardi in 1965, [2] where he described a triad of spasms in flexion, callosal agenesis, and ocular abnormalities. The syndrome is lethal in males and therefore is restricted to females. The exception is boys with XXY chromosome constitution allowing heterozygous expression of the gene as in the female. The disorder is characterized by a wide range of central nervous system (CNS) anomalies that are seen on brain imaging.[3]

CNS abnormalities include agenesis of the corpus callosum; and other CNS malformations such as ependymal cysts, choroid plexus papillomas, cortical migration abnormalities, hydrocephaly, porencephaly, cerebellar agenesis, and heterotopias.[4] Pathognomonic chorioretinal lacunae are multiple, rounded, unpigmented, and yellowish white lesions that are seen in the eye. Other ocular manifestations include optic disk coloboma, microphthalmia, and ring like pigment deposits surrounding a colobomatous papilla. A variable range of neurological symptoms and signs are often present in children with disease. The most frequent is hemiparesis or hemiplegia often on the side where the spasms predominate. A degree of microcephaly may develop but the head circumference is normal at birth. The course and outcome of AS are extremely severe. The estimated survival rate is 76% at 6 years and 40% at 14 years of age. The overall disability is usually quite marked, although variability between patients does exist.

Our child had the full spectrum of anomalies seen in Aicardi syndrome. She presented with infantile spasm, microcephaly and was found to have chorioretinal lacunae and optic disc coloboma. Her electroencephalogram showed burst suppression pattern on the left side with multifocal spike and wave discharges on the other side. Her brain MRI Showed a wide range of anomalies as described above. Currently, her seizures are controlled with vigabatrin and phenobarbitone but she has severe psychomotor delay.

References

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4. Barkovich AJ, Norman D. Anomalies of the corpus callosum: correlation with further anomalies of the brain. AJR Am J Roentgenol 1988 Jul;151(1):171-179.