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LETTER TO THE EDITOR

Aicardi Syndrome in Monozygotic Twins

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Aicardi syndrome is a rare neuro-ophthalmologic entity. It affects females only with rare exceptions. An X-linked genetic mutation being lethal to males has been proposed as a mechanism of heredity.

Two monozygotic, monoamniotic, monochorionic girls were born prematurely at 27 weeks of gestational age by emergency cesarean section.

CASE 1
The first girl’s weight 800 grams. Apgar was 8–9. At 31 weeks of post gestational age (PGA), an ophthalmologic exam was performed as a routine ROP evaluation. At that time, zone II, stage 0 ROP was noted without plus disease. Bilateral multiple chorioretinal lacunae were seen. TORCH work up was performed with negative results. ROP follow up was performed weekly until complete development of retinal vasculature. She was discharged from the hospital at 41 weeks of PGA. A month later, cerebral MRI was performed showing partial agenesis of the corpus callosum, ventricular dilation, cortical atrophy and dysplasia. She developed seizures at the age of 3 months.

CASE 2
The second child was born weighing 1000 grams. Apgar was 9–10. At 31 weeks of PGA, an ophthalmologic exam was performed as a routine ROP evaluation. A zone II stage 0 ROP was noted without plus disease, with bilateral multiple chorioretinal lacunae. ROP follow up was performed weekly and at PGA 40 weeks, zone II Stage 3 with plus disease was diagnosed in the left eye. Laser treatment was performed on the avascular peripheral retina avoiding areas of chorioretinal lacunae. Complete resolution of neovascularization was noted 2 weeks post laser.

She was discharged from the hospital at 43 weeks of PGA. A month later cerebral MRI was performed showing partial agenesis of the corpus callosum, bilateral supratentorial ventriculo-dilation, predominantly affecting the occipital horns and rectification of the frontal horns and generalized lissencephalic dysplasia. Subarachnoidal cortical spaces were broadened and the peritroncal cisterna was widened. She developed seizures at the age of 6 months. Like her twin, the patient did not have coloboma of the optic disc, microphthalmia, remnants of fetal pupillary membrane or of primary vitreous and thoracic X-rays did not show bones abnormalities. The patients did not have any other family members affected, and there were no other maternal siblings. The father had 3 more daughters and one son with two different mothers. They are reportedly normal, although unavailable for examination.

Aicardi syndrome is diagnosed in most patients by the classic triad of infantile spasms, chorioretinal lacunae and agenesis of the corpus callosum. In rare cases, one of the features may be missing, and the diagnosis can be made if two or more of the new criteria are present.

A gene responsible for Aicardi syndrome has not been identified. Some authors believe that the abnormal gene is located at the Xp-22.3 locus. The disease is not hereditary. The current hypothesis is that there is a de novo dominant X-linked genetic mutation causing disease only in females and being lethal to males. Review of the literature shows that two cases of monozygotic twins have been reported. In both cases only one of the girls developed Aicardi syndrome. Aicardi syndrome has been reported in two sisters and in a girl whose mother had suffered three miscarriages. The proposed explanation in both reports was that of a germinal mutation.

We are reporting what we believe is the first case of monozygotic twins with Aicardi syndrome. Aicardi syndrome was diagnosed because both girls developed all classic features of the syndrome. Our case report supports the theory of a genetic etiology of the disease. It is possible that a de novo mutation might have occurred early in the zygotic stage. A germinal mutation cannot be excluded given the monozygotic nature of the patients. Our patients were born prematurely. One patient developed threshold ROP in one eye in areas not affected by chorioretinal lacunae, and laser treatment was performed successfully avoiding those areas. To the best of our
knowledge this is also the first report of threshold ROP in Aicardi syndrome.

REFERENCES


