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AJNR Am J Neuroradiol 1999, 20 (6) 1148-1150
<http://www.ajnr.org/content/20/6/1148>

This information is current as
of April 18, 2024.

Case Report

Juvenile Retinoschisis: Imaging Findings

Shoaib S. Lateef, Suresh K. Mukherji, Mauricio Castillo, and David K. Wallace

Summary: We present the CT and B-scan sonographic findings in an infant with juvenile retinoschisis, a rare hereditary eye disease, which usually follows an X-linked recessive inheritance pattern.

Juvenile retinoschisis is an uncommon, inherited disease with variable degrees of severity that results in splitting of the superficial retinal layer (1). Its pathogenesis is unknown, but histopathologic and electrophysiological studies have suggested a defect in the Müller cell (1). Genetic linkage studies have localized the responsible gene to the p22 region of the X chromosome, with no evidence of genetic heterogeneity (1). We present the sonographic and CT findings in a patient with juvenile retinoschisis.

Case Report

A 5-month-old Asian-American male was noted by his primary care physician to have an abnormal red reflex of the right eye. His parents had not noticed any visual problems, and his medical and family histories were unremarkable. There was no physical evidence of trauma. On physical examination, he did not fix or follow with the right eye, but he fixed and followed well with the left eye. The pupils, ocular alignment and motility, and the anterior segments of both eyes were normal. Retinal examination showed a bullous elevation that obscured the right macula and optic nerve, and a smaller but similar inferotemporal elevation of the retina in the left eye. The differential diagnosis, based on the initial ophthalmoscopic examination, included Coat disease, bilateral retinoblastoma, familial exudative vitreoretinopathy, and retinoschisis.

Because of the retinal elevation, a noncontrast CT study of the orbits was obtained to look for a calcified mass that would be indicative of retinoblastoma (Fig 1A and B). The study revealed no evidence of a calcified mass; however, there were areas of increased attenuation present in the inferolateral quadrant of both globes that were thought to represent blood or proteinaceous fluid. The differential diagnosis based on the CT findings included Coat disease or retinal hemorrhage. At B-scan sonography, the right globe showed a complex cystic retinal mass that contained a fluid-fluid level. A similar but smaller complex cystic retinal mass was present in the left globe (Fig 1C and D).

Received January 4, 1999; accepted after revision February 17.

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Fundoscopy examination of the right eye, with the patient under general anesthesia, revealed a bullous, blood-filled cavity involving the inferior two thirds of the retina, which included the macula. The left eye had a similar elevated blood-filled cavity involving only the inferotemporal retina. The area of elevation did not extend to the ora serrata, and there were multiple cystic spaces in the central macula. These findings were consistent with bilateral juvenile retinoschisis.

Discussion

Juvenile retinoschisis involves a splitting of the sensory retina, with cyst formation between the two resultant layers (2). Histologically, the disorder is characterized by coalescent cysts, which are located primarily in the outer plexiform and adjacent nuclear layers of the retina (2). Nonunion of the retinal layers results in a retinoschisis cavity (2). This produces a translucent, veil-like membrane extending into the vitreous. The cyst is typically located in the inferotemporal quadrant of the globe (2). Extension of retinoschisis may result in destruction of the retina. Foveal schistasis is characteristic of juvenile retinoschisis, occurring in 98% to 100% of patients (2). Although macular changes are present in all affected patients, some investigators have found the typical foveal schistasis to be present in only 70% of patients (2).

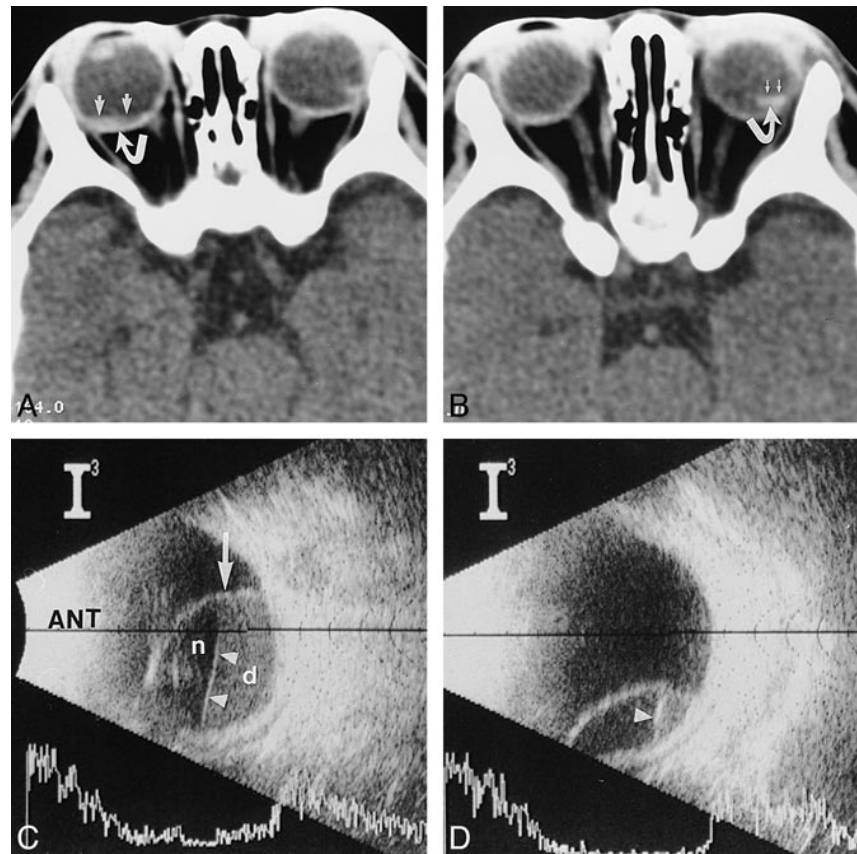
Histologic examination reveals that the cystic contents are filled with an amorphous proteinaceous material that is hyaluronidase negative (2). Electron microscopy suggests that this material contains filaments of similar periodicity to that of the vitreous and that it blends with the basement membrane of Müller cells (6). It can therefore be assumed that these intraretinal filaments are produced by defective Müller cells and that their extracellular accumulation leads to retinal splitting and subsequent schistasis (6). The areas of increased attenuation seen on CT scans most likely represent the regions of increased protein content that may have resulted from hemorrhage into the cysts. These areas probably correspond to the areas of high echogenicity identified within the cysts on B-scan sonograms.

Juvenile retinoschisis is a rare disorder, whose exact prevalence is unknown; but it has been found worldwide in white, black, and Asian populations (3). It usually manifests during school age, when reading difficulties become evident. Less commonly, the disorder may present in early infancy, as was the case in our patient, with such signs as strabismus, nystagmus, and bilateral highly elevated bul-

FIG 1. 5-month-old boy with juvenile retinoschisis.

A and B, Axial noncontrast CT scan through the inferior aspect of the orbits shows an area of increased attenuation that appears to be layering along the posterior aspect of the right globe (*curved arrow, A*) and in the lateral (temporal) aspect of the left globe (*curved arrow, B*). This was thought to represent blood or other proteinaceous fluid. The nondependent (anterior) surface of this collection is flat and therefore suggestive of a fluid-fluid level (*straight arrows*).

C and D, B-scan sonograms of the right (C) and left (D) globes. A complex cystic mass in the right globe abuts the retina and contains a fluid-fluid level (*arrowheads, A*). The high echotexture component of the mass present in the dependent portion (*d*) probably represents highly proteinaceous fluid or layering blood products and corresponds to the area of increased attenuation seen on the CT scans. The lower echotexture component in the nondependent portion (*n*) is probably composed of fluid with low protein content (*ANT* indicates anterior). The B-scan sonogram of the left globe (D) shows a similar but smaller mass (*arrowhead indicates fluid-fluid level*).



lous cavities, often with hemorrhage in the cavity or in the vitreous.

The disease is slowly progressive, and most patients retain relatively good vision until the fifth or sixth decades of life, when macular atrophy develops (4). On average, visual acuity is around 20/60 at age 20, gradually declining to 20/200 by age 60, usually as a result of macular changes (4). The most serious vision-threatening complications associated with juvenile retinoschisis are retinal detachment and vitreous hemorrhage. Retinal detachments have been reported to occur in 20% of cases (2); vitreous hemorrhage, which is believed to result from rupture of unsupported retinal vessels or, rarely, from neovascularization, has been reported in up to 40% of patients (4).

The mechanism of disease in congenital juvenile retinoschisis involves inherently weak Müller cell pillars (5). The Müller cell, the principal glial cell of the retina, is central to the migration and organization of other retinal cells during development. A gene defect in the Müller cell could account for the structural and physiological abnormalities found in juvenile retinoschisis (5). The inheritance pattern of this Müller cell defect may be X-linked recessive, autosomal dominant, or autosomal recessive (6). The gene for the X-linked form of the disease has been localized to Xp22, and there is no evidence of genetic heterogeneity (7). Localization of the gene means that positional cloning and char-

acterization, as well as confirmation of diagnosis and genetic counseling, may soon be feasible.

There is no known cure for juvenile retinoschisis. A correct and early diagnosis is important for accurate prognosis, genetic counseling, and treatment of children with visual handicaps. Strabismus, refractive errors, and amblyopia should be corrected to maximize visual potential (3). Panretinal photocoagulation has been used successfully to treat disk and peripheral neovascularization (3). Vitreous hemorrhage usually clears spontaneously, although vitrectomy may be required to reduce the risk of amblyopia (3).

Conclusion

The CT findings in our patient were helpful for delineating the extent of the retinoschisis abnormality and for excluding retinoblastoma. Although CT findings are not specific for retinoschisis, the presence of a fluid-fluid level containing high-attenuation fluid that layers dependently may be suggestive of the diagnosis. When combined with the results of B-scan sonography and clinical examination, CT findings are beneficial in reaching an early diagnosis.

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