Coats Disease and VATER Association in a 5-Year-Old Boy

Coats disease is an uncommon exudative retinopathy of unknown origin that may cause blindness. We report the unusual occurrence of Coats disease in a 5-year-old boy with multiple congenital abnormalities due to VATER association. The possibility of a genetic basis for some cases of Coats disease is discussed.

The VATER association comprises congenital defects including vertebral defects, imperforate anus, tracheoesophageal fistula, and radial and renal dysplasia. The underlying cause of VATER association may be a severe embryonic insult during the simultaneous development of the organ systems. Patients with VATER association often require multiple surgical procedures and extensive rehabilitation. We report a unique case of exudative retinopathy (Coats disease) in a case of VATER association.

Report of a Case. A child with a normal prenatal ultrasound at 20 weeks gestation and a negative TORCH (toxoplasmosis, other infections, rubella, cytomegalovirus infection, and herpes simplex) panel was born after an otherwise uncomplicated pregnancy by normal spontaneous vaginal delivery at 33 weeks gestational age. The results of a physical examination at birth revealed an imperforate anus and polydactyly with an extra digit lateral to the right thumb. The results of a radiological examination showed communicating hydrocephalus with grade II bilateral renal reflux and hemivertebra with severe scoliosis (Figure 1A). Corrective surgical procedures included digit amputation, colostomy, and anoplasty. There was no family history of eye or genetic defects, and karyotyping was normal in the child and his parents.

At 5 years of age, the patient was referred for left eye leukocoria. Visual acuity was 20/20 OD and 1/60 OS. There was total exudative retinal detachment with associated tortuous vessels with multiple aneurysmal dilatations compatible with Coats disease, stage 3B (Figure 1B). The lower 2 quadrants of the retina were preferentially affected. There was no evidence of associated anterior chamber or vitreous inflammation. The findings of the retinal examination of the fellow eye, including the periphery area, were normal. Further examination of old photographs showed the patient had had leukocoria since the age of 6 months. The results of B-scan ultrasonography revealed retinal detachment with no calcification or mass. This was confirmed by a computed tomography scan of the orbits. Intraocular pressure was normal with no evidence of neovascularization of the iris. Cryotherapy was offered to the patient to preserve vision and prevent further complications, including neovascular glaucoma, a painful blind eye, and a possible need for enucleation. The parents finally opted for conservative treatment.

Comment. A number of patients with VATER association have been described as having eye defects. The most common associations are coloboma and microphthalmos. However, the retina and posterior segment are seldom involved, and Coats disease was hitherto unreported. The term Coats disease refers to idiopathic retinal telangiectasia with intraocular or subretinal exudation and without appreciable signs of retinal or vitreous traction. The diagnosis of Coats disease is one of exclusion after careful workup and examination, especially with regard to retinoblastoma, retinopathy of prematurity, retinal capillary angiomatosis, and toxocara, all of which were not implicated in our case. Reported associations include renal-retinal abnormalities and retinal disorders. Familial forms exist, but no specific gene defect is known. In the largest reported series, the median

A, Scout film of a computed tomography scan showing absent hemivertebrae (arrows) and severe scoliosis typical of VATER association. B, Indirect funduscopy photograph showing severe exudative retinopathy with retinal detachment.
age of diagnosis was 5 years with a male predominance (76%), the majority with unilateral disease (95%). Aggressive management for early cases, including laser photoagulation, cryotherapy, and drainage of subretinal exudates using pars plana vitrectomy techniques, may prevent neovascular glaucoma and a painful blind eye.

Because the definitive genetic defects for both VATER association and Coats disease in humans are unknown, we can only speculate about the possible common genetic link. Recently, an adriamycin-induced rat model of the VATER association, with defects in the hedgehog gene pathway, has been developed. The secreted glycoprotein, sonic hedgehog (SHH), acts as an endodermal signal that controls gut and lung patterning. Interestingly, the SHH protein is also secreted by retinal ganglion cells to help optic disc astrocyte precursor cells to guide retinal axon growth, and to convert optic stalk neuroepithelial cells into pigmented cells. Both VATER and retinal defects can be produced by targeted SHH mutations. Ultrastructural retinal cell abnormalities are not infrequently found in pathologically examined eyes of patients with Coats disease. Nevertheless, it remains to be elucidated whether any aberrations in these pathways may explain the unusual clustering of specific defects in our case.

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**Idiopathic Isolated Abscess in an Extraocular Muscle in a Child**

An idiopathic muscle abscess is a rare condition that should be considered in the differential diagnosis of painful limitations of eye movements and double vision in children.

We herein present the case of an idiopathic muscle abscess in an 11-year-old girl. Myositis was initially suspected, but the magnetic resonance imaging (MRI) scan showed typical signs of an abscess. Antibiotic therapy led to a rapid and complete improvement of all symptoms. To our knowledge, this is the first report of an idiopathic isolated abscess in an extraocular muscle in a child.

**Figure 1.** Patient photographs before (A) and after (B) treatment.

**Figure 2.** Oblique sagittal (A) and axial (B) T1-weighted contrast-enhanced spin-echo magnetic resonance imaging (MRI) scans. Diffuse swelling of the left superior rectus muscle is seen surrounding an abscess (diameter, 8 mm), with central cavitation and rim of contrast enhancement of about 3 mm surrounded by a diffuse swelling of the left superior rectus muscle (Figure 2A and B).