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A Case of Congenital Mydriasis Suspected Multisystemic Smooth Muscle Dysfunction Syndrome

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Abstract | **Case Report**

We present a case of congenital mydriasis in a 6-year-old boy who had a history of patent ductus arteriosus. Anterior segment examination revealed mydriasis in both eyes. Fundus examination revealed retinal arteriolar tortuosity in both eyes. Congenital mydriasis is extremely rare, and the incidence of extreme patent ductus arteriosus is low. We discuss congenital mydriasis as a separate entity and in combination with heart diseases.

Keywords: Congenital mydriasis, retinal arteriolar tortuosity, patent ductus arteriosus.

INTRODUCTION

Congenital mydriasis, sometimes referred to as congenital fixed dilated pupil, is a rare condition [1, 2]. Multisystemic smooth muscle dysfunction syndrome is characterized by dysfunction of smooth muscle throughout the body leading to congenital fixed dilated pupils with defective accommodation, persistent ductus arteriosus, aortic and cerebrovascular disease, hypotonic bladder, intestinal hypoperistalsis and pulmonary hypertension [3, 4]. It is due to a p.R179H mutation in the *ACTA2* gene, chromosome 10q23.3 that is predicted to express a mutant α-actin in smooth muscles [3, 4]. Herein, we report a 6-year-old boy with congenital mydriasis.

CASE REPORT

A 6-year-old boy presented with the complaint of diminished vision in his both eyes. He had a history of patent ductus arteriosus. On examination, his best corrected visual acuity was 0.4 in both eyes. Anterior segment examination revealed mydriasis in both eyes (Figure 1A, B). The pupils measured 6.5 mm and were nonreactive to light, accommodation. Fundus examination revealed retinal arteriolar tortuosity in both eyes (Figure 1C, D). We diagnosed our patient with congenital mydriasis.

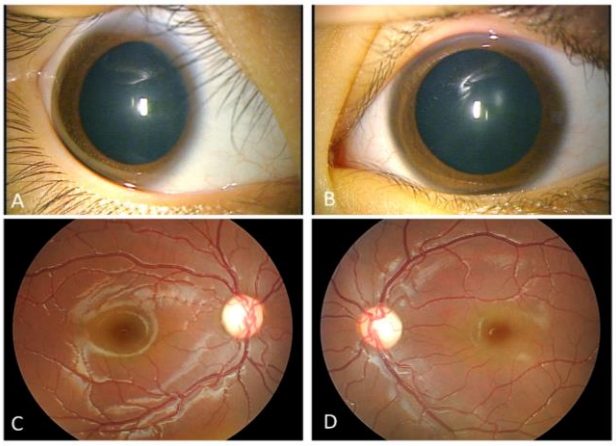

Figure 1: Slit-lamp and fundus photographs of the right (A, C) and the left (B, D) eyes

Note mydriasis and retinal arteriolar tortuosity in both eyes.

Cycloplegic retinoscopy revealed a refractive error of a +5.00D in the right eye and +4.50D in the left eye. Glasses with full optical correction were prescribed. Because of the iris features and retinal vascular tortuosity associated with patent ductus arteriosus, we speculate multisystemic smooth muscle dysfunction. However, genetic testing of the *ACTA2* gene could not performed.

DISCUSSION

Congenital mydriasis is extremely rare, and the incidence of extreme patent ductus arteriosus is low [1, 5-7]. The mechanism behind the link between these two very rare diseases is not known.

In 2010, Milewicz *et al*., [3] reported 7 unrelated patients with a de novo missense mutation in the *ACTA2* gene (p.R179H) and multisystemic smooth muscle dysfunction syndrome, including aortic and cerebrovascular disease, patent ductus arteriosus, and congenital fixed dilated pupils. Moller *et al*., [5] reported the ophthalmic features of 3 patients with multisystemic smooth muscle dysfunction syndrome. They all presented with normal visual acuity, dilated nonreactive pupils, impaired accommodation, and retinal vascular tortuosity. Mutations in *ACTA2* gene lead to a multisystemic smooth muscle dysfunction syndrome that causes vascular disease, congenital mydriasis, and variable presentation of urinary and gastrointestinal problems [3-10]. Among the vascular manifestations are aortic aneurysms and dissections, patent ductus arteriosus and cerebrovascular disease [3-10]. Retinal vessel tortuosity is usually regarded an occasional finding, but when combined with persistent ductus arteriosus and the extremely rare occurrence of congenital mydriasis, immediate attention should be drawn to the multi systemic smooth muscle cell dysfunction syndrome due to an *ACTA2* missense mutation leading to the production of mutant cellular α-actin.

CONCLUSION

Fixed dilated pupils were extremely rare condition and should alert pediatricians and ophthalmologists to the possibility of the coexistence of systemic life threatening disorders, including multisystemic smooth muscle dysfunction syndrome. Genetic testing is essential in evaluating this patient population.

Conflicts of Interest: The authors have no financial or proprietary interests related to this paper.

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