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Bilateral congenital mydriasis

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Abstract

A single case of bilateral congenital mydriasis is described. A review of the literature is presented and possible modes of inheritance are discussed.

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Congenital mydriasis is an extremely rare ophthalmic condition. By definition the fixed and dilated pupils are present from birth and occur in an otherwise normal seeing eye.[1,2] All previously reported cases are bilateral and have occurred in females. We report the clinical findings of a 2½-year-old girl who has congenital mydriasis and developmental delay. The condition of congenital mydriasis should be considered separate from aniridia both in clinical appearance and visual potential.

Case report

A 2½-year-old girl was referred with the diagnosis of general developmental delay and bilateral congenital, fixed, dilated pupils. The girl was born at term after an uneventful pregnancy and delivery. From birth the mother noted dilated, unreactive pupils. The child's main problem was developmental delay. The infant was floppy and was late in achieving her motor milestones. Visually the child also seemed delayed. She did not smile to a visual stimulus until 6 months of age. The parents became concerned at this stage and consulted an ophthalmologist who ordered an ERG which was normal.

A fine horizontal nystagmus was present during infancy which has diminished with time such that no nystagmus was detected at age 2½ years. Concurrent with the reduction in nystagmus has been an improvement in her visual function.

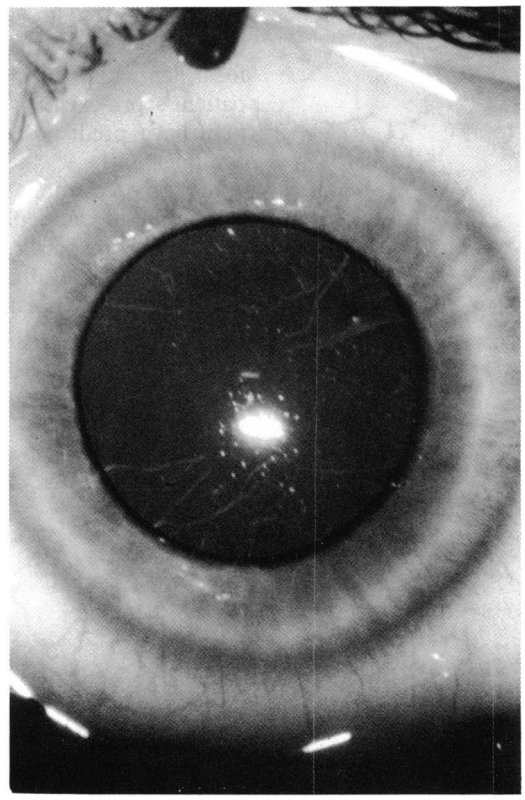

Figure 1 Rim of iris seen with pupillary ruff. Fine pupillary membrane present.

There is no family history of note, no consanguinity of the parents, and the child has a healthy 4-year-old sister. At the time of the examination at age 2½, the mother felt her child was seeing normally. The parents thought that the child was light sensitive, though this was not borne out when the examination took place. The general examination was normal. (Formal assessment of motor and social skills by Griffith's

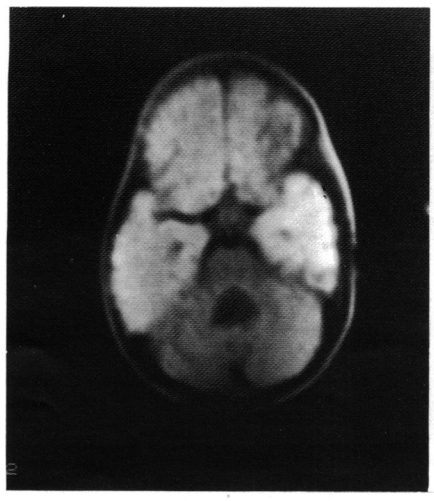

Fig 2A

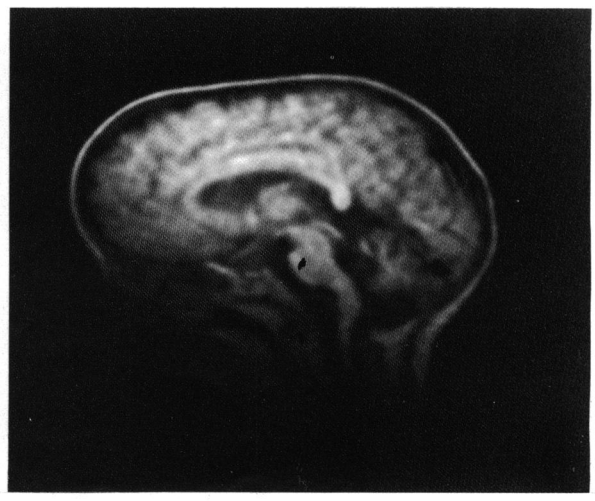

Fig 2B

Figure 2 Transverse (A) and sagittal (B) spin-echo (SE 544/44) images of the brain showing an enlarged fourth ventricle, atrophic vermis of the cerebellum, and normal aqueduct and midbrain. The left lateral ventricle was slightly dilated on high images (not shown).

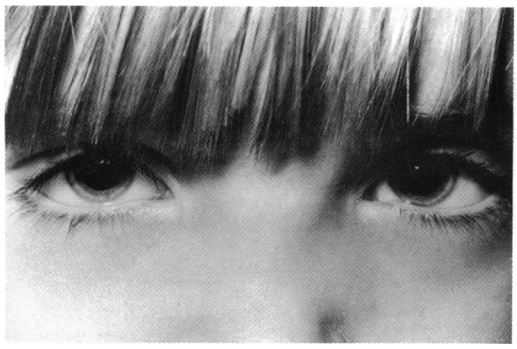

Figure 3 Pupil size before pilocarpine.

Figure 4 Pupil size 60 minutes after pilocarpine.

scoring revealed a level achieved of 15 months when the child was aged 30 months.) The pupils were dilated and unreactive to light or the near reflex. The pupil diameter was 7 mm and round and symmetrical. The irises themselves were grossly normal in appearance. There was a reasonable rim of iris and a fine, persistent pupillary membrane (Fig 1). The cornea, lens, vitreous, optic nerve, macula, and peripheral retina were entirely normal on examination. Corneal diameter was 12·5 mm left and right, intraocular pressures, right 9 mm Hg, left 8 mm Hg. An examination under anaesthetic was performed; the angle architecture was normal on gonioscopy. A patchy transillumination defect of the peripheral iris was noted on examination with the operating microscope.

The child was extensively investigated and urinalysis was normal. There was no haematuria; urea and electrolytes, osmolarity, urine amino acids, organic acids, blood gases, plasma lactate, and chromosome studies were all normal. A pattern evoked ERG was performed which was normal, and occipital VEPs to pattern reversal stimulation were evident from each eye for medium sized checks. CT scan and MRI brain scan did show an enlarged fourth ventricle, some hypoplasia of the vermis of the cerebellum, and a normal midbrain (Fig 2). Attempts were made to constrict and dilate the iris with pilocarpine and atropine respectively; however, photographically, no change was discernible (Figs 3 and 4).

Discussion

Eleven cases of bilateral congenital mydriasis have been reported previously in the literature; all 11 are female. The first two cases reported by White and Fulton[1] occurred in monozygotic twins and the eye abnormality was described as large, irregular pupils which reacted only slightly to light. The authors suggested that the abnormality was due to congenital absence of the sphincter iridis. Although not examined, the twins' mother was also said to have a similar iris anomaly suggesting a dominant inheritance.

Caccamise and Townes[2] reported a single case of congenital mydriasis and on the patient's evidence identified seven other members of the family with similar findings. No males were found to have congenital mydriasis. The authors agreed with White and Fulton that the likely mode of inheritance was autosomal dominance. The authors pointed out that some form of sex limitation such as occurs in incontinentia pigmenti either X chromosome linked or autosomal dominant with non-viability of males could not be excluded. Our case, also female, strengthens the sex limitation theory for the mode of inheritance. Caccamise and Townes found slight pupillary constriction to light and pilocarpine, and dilatation to phenylephrine 10%. They agreed that the iris sphincter and dilator were present. We were unable to identify any pupil constriction to pilocarpine (Figs 3 and 4).

Our patient was referred by non-ophthalmologists with the presumptive diagnosis of aniridia. The child has none of the ocular features of aniridia and the iris structure itself appears normal albeit in a fixed, dilated state. Sporadic cases of aniridia are associated with Wilm's tumour and aniridic eyes typically have poor vision. There is no evidence of these associations in congenital mydriasis and the distinction between the two conditions should be clear.

Summary

A case of bilateral congenital mydriasis is presented in a 2½ year-old child with developmental delay. All previously reported cases have been female. Our case, also female, further supports the theory of a form of sex limitation with non-viability of males as the mode of autosomal dominant inheritance. The visual potential in this condition is normal and a clear distinction from aniridia should be made.

1 White BV, Fulton MN. A rare pupillary defect inherited by identical twins. *J Hered* 1937; **28**: 177–9.
2 Caccamise WC, Townes PL. Bilateral congenital mydriasis. *Am J Ophthalmol* 1976; **81**: 515–7.