ABSTRACT

Spectrum of congenital and developmental anomalies of eye

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Purpose: The purpose of this study was to determine the type and frequency of congenital/ developmental defects and to gather baseline data for future studies on congenital/ developmental anomalies of eye.

Design: Retrospective and prospective case series

Participants and methods: The study was conducted at Al-Shifa Trust Eye Hospital Rawalpindi over a period of 3 years in the Paediatric Ophthalmology Department which included recording of visual acuity, complete anterior and posterior segment examination and cycloplegic refraction of 514 patients that formed the study group.

Results: 45.3% of the children had no visual impairment, while 54.7% had severe visual impairment/blindness. The most frequent anomaly seen was cataract (23%), followed by nasolacrimal duct obstruction (20%). Congenital ptosis, followed by blepharophimosis was important lid anomalies. Among the retinal disorders (15% of the total), pigmentary retinal degenerations contributed to 44% while albinism and maculopathies were the other important anomalies. Congenital anomalies of the optic nerve, cornea and defects involving the whole globe were found less frequently. Consanguinity was positive in 56% of the cases. A positive family history was obtained in 10% of the cases. 77.5% of the children were amenable to treatment at the time of presentation, while 22.5% could not be offered any treatment. Overall, more than half of the children (51.2%) underwent some sort of surgery. Spectacles, low vision aids and conservative management were offered as other mode of therapy.

Conclusion: In Pakistan there is significant prevalence of congenital anomalies. Consanguinity is one of the major contributory factors for hereditary and congenital eye diseases. Public health education about the problems of consanguinity and community awareness about availability of genetic counseling services needs to be promoted. Al-Shifa Journal of Ophthalmology 2007; 3(2): 56-60 © Al-Shifa Trust Eye Hospital, Rawalpindi, Pakistan.