Ocular Genetics and Down Syndrome

These are exciting times for pediatric ophthalmologists involved in the care of children with ocular findings resulting from genetic defects. The rapidly expanding field of molecular genetics has resulted in more accurate diagnosis and more precise prediction of inheritance. Therapeutic measures are on the horizon. In Leber’s congenital amaurosis, a mutation in the gene RPE65 results in a deficiency of 11-cis retinal so that rod photoreceptor cells are unable to respond to light and progressive degeneration of cone photoreceptor cells follows, resulting in the loss of cone-mediated vision. In a small but exciting study, the subretinal injection of recombinant adeno-associated virus RPE 65 resulted in evidence of efficacy in improved visual function in humans.¹

Excellence in the practice of pediatric ophthalmology demands a thorough knowledge of the myriad genetic disorders that manifest ocular signs and symptoms. The ability to perform an evaluation, reach a diagnosis, and formulate a treatment plan in this often challenging group of patients defines what pediatric ophthalmology is all about.

Down syndrome remains the most common genetic condition presenting to pediatric ophthalmologists. Two articles in this issue, one concentrating on the incidence of refractive errors in this population, review the ocular findings in these patients. In their review, Creavin and Brown determined that pediatricians were good at identifying ptosis, keratoconus, congenital glaucoma, detached retina, and nystagmus, for which their findings were the same or almost the same as those of the ophthalmologist. However, pediatricians were less proficient at identifying strabismus, refractive error, lacrimal disease, and blepharitis. Their results suggested that a non-specialist examination of children with Down syndrome, in isolation, may miss approximately 20% of ophthalmic disorders. The authors suggest pediatric ophthalmologic monitoring of all children with Down syndrome. This is certainly a reasonable approach to insure optimal care for these children.

REFERENCE


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