Correspondence

Usher's syndrome

Sir, Fishman et al. attempt to correlate age-related loss of central visual acuity in the Usher syndrome with 'quantitative' estimates of 'contributing factors' such as foveal lesions and cataracts in hopes of generating data for the purposes of counselling patients 'as to the potential for visual loss with age'. These authors may have overlooked the most useful and reliable predictor of age-related visual loss in the Usher syndrome, namely, the degree of congenital neurosensory hearing loss. Several investigators have suggested that the Usher syndrome is genetically heterogeneous, with approximately 90% of patients having profound congenital deafness, vestibular dysfunction, and onset of retinal dystrophy before age 10 years, and 10% having some residual hearing with normal vestibular function and onset of retinal dystrophy after adolescence. Although both forms are inherited in an autosomal recessive way, they appear to be two genetically distinct entities that show little overlap between families.

Degree of sensorineural hearing loss and age of onset of symptoms of retinal dystrophy would appear to be better predictors of age-related visual acuity than foveal lesions, since there was not an invariable correlation between decreased visual acuity and foveal lesions in the study of Fishman et al., nor was there an attempt to determine the predictive value of a given foveal appearance for visual acuity over time.

Prospective studies to define the natural history of genetic disorders such as the Usher syndrome are badly needed. A 'sampling' of larger numbers of patients will not clarify the natural history unless all possible factors that may predict rate of age-related visual loss are studied.

ROBERTA A. PAGON

Division of Medical Genetics,
Children's Orthopedic Hospital
and Medical Center,
4800 Sand Point Way NE,
Seattle, Washington 98105, USA.

References
