Chapter 14

Wagner Syndrome and Stickler Syndrome

The characteristic features of Wagner syndrome include myopia, an optically empty vitreous, preretinal membranes, perivascular retinal pigment epithelial changes, vascular sheathing, cataracts, and atrophy of the choriocapillaris. The cataracts typically appear as patients approach their 40th birthday. The vitreous bands are broad and insert into the peripheral retina; and there is liquefaction of the vitreous. It is autosomal dominant and usually present bilaterally. It is noteworthy that many physicians consider Wagner syndrome to be a mild form of Stickler syndrome. Wagner syndrome patients, however, do not have systemic changes, whereas Stickler syndrome patients have systemic manifestations. The former patients do not suffer retinal detachments, whereas more than 50% of the Stickler syndrome patients develop a retinal detachment.

Visual acuity is normal if both the posterior pole and lens are normal. Color vision is usually normal. Although the electroretinogram is normal early in the disease, as time progresses it becomes more and more abnormal.

The addition of systemic manifestations to Wagner syndrome encompasses the entity known as Stickler syndrome, or hereditary progressive arthroophthalmopathy. As with Wagner syndrome, it is a progressive hereditary disorder with an autosomal dominant pattern of inheritance. The systemic aspects of this disease involve the facial and skeletal systems. The ocular signs of the disease are similar to those seen with Wagner syndrome. The myopia (usually more than 10 diopters), retinal breaks, cataracts, and glaucoma appear to be progressive in this disorder. The prognosis for retinal detachment surgery is usually unfavorable. It is estimated that at least 50% of these patients experience a detachment during their lifetime. These patients also seem to have a high incidence of hearing loss. They have flattened facies, a cleft palate, and arthropathy. Stickler syndrome patients can be categorized into four subgroups, as was done by Lisch in 1983: (1) those with facial abnormalities and a cleft palate; (2) those with an Eskimo-like face; (3) those with hyperextensibility of joints and a cleft palate; and (4) those with progressive arthroophthalmopathy.
Color vision is unaffected in these patients. The visual fields may demonstrate constriction. Electrophysiological studies in Stickler syndrome patients are similar to those seen in patients with Wagner syndrome, that is, subnormal. Dark adaptation may be normal or abnormal. It should be remembered that retinal detachments and an oral cavity abnormality bespeaks of a variant of Stickler syndrome commonly known as “clefting syndromes.”

Selected Reading