SAG, also known as *S-antigen* or *S-arrestin*, encodes a rod photoreceptor protein involved in the recovery phase of light transduction. It is present in the retina and pineal gland and plays an inhibitory role in the activated phototransduction cascade [1, 2]. Mutations are responsible for Oguchi disease and for retinitis pigmentosa (RP).

SAG mutations are responsible for the majority of cases of Oguchi disease in the Japanese population and have an autosomal recessive pattern of inheritance. The most frequent mutation in this population is c.926delA; p.N309Tfs*12 (previously reported as 1147delA) [2, 3]. Hayashi et al. (2010) reported that 5 out of 6 Japanese patients with Oguchi disease are homozygous for the c.926delA mutation [3]. Oguchi is a form of congenital stationary night blindness (CSNB) where the only symptom is generally a childhood-onset of nyctalopia [4]. Visual acuity, visual field, and color vision are usually all within normal range. A characteristic feature of Oguchi is the diffuse golden yellow or gray discoloration of the fundus, which disappears after prolonged dark-adaptation and reappears almost immediately upon re-exposure to light (called the Mizuo-Nakamura phenomenon) [2–4]. Full-field electroretinogram (ERG) is non-recordable after 30 minutes of dark adaptation, but cone and flicker ERG recordings are essentially normal. Combined rod-cone ERG responses show a significantly reduced a-wave, a non-detectable b-wave, and well-preserved oscillatory potentials [3, 4].

SAG mutations have variable expressivity. Some patients have Oguchi disease only [2] and some manifest symptoms RP along with the Mizuo-Nakamura phenomenon [4]. There is a report of a patient with autosomal recessive RP with a

homozygous c.926delA mutation in *SAG* who has pigmentary retinal degeneration and retinal pigment epithelium (RPE) atrophy along the vascular arcades. This patient has constricted visual fields and paracentral visual field defects and also exhibits the Mizuo-Nakamura phenomenon [3].

More recently, Sullivan et al. described a dominantly-acting mutation in *SAG* (c.440G>T; p.Cys147Phe) that is responsible for 36% of the autosomal dominant retinitis pigmentosa in Hispanic patients in the United States. All these patients demonstrated classic fundus features of retinitis pigmentosa with a rod-cone pattern of ERG loss; none demonstrated the Mizuo-Nakamura phenomenon [5].

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