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GNAT1 (Guanine nucleotide-binding protein, alpha-transducing activity polypeptide 1) encodes the alpha subunit of rod transducin. Transducin is the G-protein involved in the phototransduction cascade that facilitates the interaction between rhodopsin and cGMP. Mutations in *GNAT1* have been shown to cause both autosomal recessive and autosomal dominant forms of congenital stationary night blindness (CSNB) and autosomal recessive rod-cone dystrophy [1, 2].

Autosomal dominant CSNB may show non-progressive night blindness from early infancy. Myopia is common, while visual acuities and color vision are typically within normal limits. GVF and fundus appearance are typically normal. The Nougaret type of CSNB, based on the pedigree of Jean Nougaret, demonstrates an electroretinogram (ERG) abnormality in which the rod a-wave is missing [2]. In contrast, a Riggs-type ERG pattern reveals an absent b-wave in response to flashes of dim light and a cone-like response to bright light in scotopic testing conditions. Light-adapted states are typically normal. Dark adaptation testing reveals reduction in rod sensitivities in most patients. Some studies have suggested that these ERG findings reflect a presynaptic defect in rod phototransduction in patients with CSNB [3].

Autosomal recessive CSNB has been reported in one study of a consanguineous family that had many affected family members. Night blindness may start from early childhood, with unaffected visual acuity and color vision. Fundus findings are typically normal, with no signs of arteriolar attenuation or bone spicule changes. ERG recordings may reveal nearly absent a- and b-waves in scotopic conditions with normal photopic parameters [1].

There are a few case reports of patients with homozygous *GNAT1* pathogenic variants associated with late-onset rod-cone dystrophy.

References

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