

## Achromatopsia via Sequential Sequencing of *CNGB3, CNGA3, GNAT2 and PDE6C* Genes -- Test #695

**Brief Description of Clinical Features:** Achromatopsia (ACHM) is a congenital cone rod dystrophy (CRD) that can be distinguished from other CRDs on the basis of primary cone involvement, stationary course, and normal fundus (Hamel Orphanet J Rare Dis 1:2-7, 2007). Two clinical types of achromatopsia, complete and incomplete, are recognized. In patients with complete achromatopsia, symptoms usually begin in infancy and include nystagmus, low visual acuity, photophobia, severe color vision defects, and selective absence of functioning cone photoreceptor cells in electroretinogram (ERG) findings. Patients with incomplete achromatopsia retain residual functioning cone cells. In addition, they have mild visual acuity and mild color vision defects. The prevalence of complete achromatopsia is 1 per 30,000 people worldwide (Michaelides et al. Br. J. Ophthalmol 88, 291–297, 2004). However, in the Micronesian atoll of Pingelap, achromatopsia affects ~ 5 % of the island population (Morton et al. Am J Hum Genet 24:277-289, 1972).

**Genetics:** Achromatopsia is a heterogeneous genetic disease that is inherited in an autosomal recessive manner. It is caused by defects in various genes that encode important elements of the cone phototransduction process. Over 130 different causative mutations were detected in the four genes listed below, accounting for ~75% of all patients with a clinical diagnosis of achromatopsia (Kohl et al. GeneReviews, 2010, [www.genetests.org](http://www.genetests.org)). See also individual gene Test Descriptions.

**Description of These Tests:** PreventionGenetics offers sequencing of each of the four genes individually, or the sequential Panel described here. These tests involve bidirectional DNA sequencing of all coding exons of the genes as well as ~50 bp of flanking-coding DNA on either side.

**Reference Sequences:**

Gene	<i>CNGB3</i>	<i>CNGA3</i>	<i>GNAT2</i>	<i>PDE6C</i>
<b>Genomic NC_</b>	000008.10	000002.11	000001.10	000010.10
<b>mRNA NM_</b>	019098.4	001298.2	005272.3	006204.3
<b>Protein NP_</b>	061971.3	001289.1	005263.1	006195.3
<b>Sensitivity</b>	~ 50%	~ 25%	~ 2%	~ 2%

**Indications for Test:** Patients with normal rod response and absence of cone response in ERG findings

**Sensitivity of Test:** Together, these four tests may detect mutations in up to 75% of all ACHM patients (see table above; Kohl et al. 2010).

**Turnaround Time:** Maximum of 50 days, although many tests are completed in 2-3 weeks.

**Specimen Requirements:** See page 4 of Requisition Form

**Prices and CPT Codes:**

CPT	Description	<i>CNGB3 Only</i>	<i>CNGA3 Only</i>	<i>GNAT2 Only</i>	<i>PDE6C Only</i>	<i>All Four Genes*</i>
<b>83890</b>	Sample Ascertainment	\$ 30 (x1)	\$ 30 (x1)	\$ 30 (x1)	\$ 30 (x1)	\$ 30 (x1)
<b>83891</b>	DNA Isolation	\$ 40 (x1)	\$ 40 (x1)	\$ 40 (x1)	\$ 40 (x1)	\$ 40 (x1)
<b>83898</b>	Amplification	\$ 290 (x18)	\$ 150 (x8)	\$ 150 (x8)	\$ 330 (x21)	\$ 930 (x55)
<b>83904</b>	Sequencing	\$ 440 (x18)	\$ 220 (x8)	\$ 220 (x8)	\$ 500 (x21)	\$ 1390 (x55)
<b>83894</b>	Separation	\$ 70 (x1)	\$ 40 (x1)	\$ 40 (x1)	\$ 80 (x1)	\$ 180 (x1)
<b>83912</b>	Interpretation/Report	\$ 110 (x1)	\$ 90 (x1)	\$ 90 (x1)	\$ 110 (x1)	\$ 160 (x1)
	<b>Totals</b>	<b>\$ 980</b>	<b>\$ 570</b>	<b>\$ 570</b>	<b>\$ 1090</b>	<b>\$ 2730</b>

\*When three genes are sequenced, the total cost will be discounted 15%.

**Accreditation:** CLIA ID #: 52D1027685 (expires 1/18/13) (CAP#: 7185561, AU ID: 1407125 expires 12/20/12)

**Contact:** Dr. Khemissa Bejaoui, [khemissa@preventiongenetics.com](mailto:khemissa@preventiongenetics.com), [www.preventiongenetics.com](http://www.preventiongenetics.com)