Albinism is a group of inherited disorders in which melanin biosynthesis is reduced or absent [1]. The lack or reduction in pigment can affect the eyes, skin and hair, or only the eyes. In addition, there are several syndromic forms of albinism in which the hypopigmented and visual phenotypes are seen in addition to other systems involvement [2].

Our Albinism Panel includes analysis of the genes listed below.

<table>
<thead>
<tr>
<th>Oculocutaneous Albinism</th>
<th>Ocular Albinism</th>
<th>Hermansky-Pudlak Syndrome</th>
<th>Chediak-Higashi Syndrome</th>
<th>Griscelli Syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>TYR</td>
<td>SLC45A2</td>
<td>HPS1</td>
<td>HPS4</td>
<td>BLOC1S3</td>
</tr>
<tr>
<td>OCA2</td>
<td>SLC24A5</td>
<td>AP3B1</td>
<td>HPS5</td>
<td>BLOC1S6</td>
</tr>
<tr>
<td>TYRP1</td>
<td>C10ORF11</td>
<td>HPS3</td>
<td></td>
<td>DTNB1</td>
</tr>
</tbody>
</table>

Oculocutaneous Albinism
Oculocutaneous albinism (OCA) is a genetically heterogeneous congenital disorder characterized by decreased or absent pigmentation in the hair, skin, and eyes. Clinical features can include varying degrees of congenital nystagmus, hypopigmentation and translucency, reduced pigmentation of the retinal pigment epithelium and foveal hypoplasia. Vision acuity is typically reduced and refractive errors, color vision impairment and photophobia also occur [3].

Ocular Albinism
Ocular albinism (OA) is characterized by nystagmus, impaired visual acuity, iris hypopigmentation with translucency, albinotic fundus, macular hypoplasia, and normally pigmented skin and hair.

Hermansky-Pudlak Syndrome
Hermansky-Pudlak syndrome (HPS) is characterized by oculocutaneous albinism, bleeding tendency, and ceroid deposition, which likely leads to deleterious lesions in lungs, heart, and other organs. Early detection and involvement of appropriate management may mitigate the risks of life-threatening complications and provide prognostic information [8].

Chediak-Higashi Syndrome
Chediak-Higashi syndrome is characterized by decreased pigmentation of hair and eyes (partial albinism), photophobia, nystagmus, large eosinophilic, peroxidase-positive inclusion bodies in the myeloblasts and promyelocytes of the bone marrow, neutropenia, abnormal susceptibility to infection, and peculiar malignant lymphoma.

Griscelli Syndrome
Griscelli syndrome is characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes.

Test methods:
Comprehensive sequence coverage of the coding regions and splice junctions of all genes in this panel is performed. Targets of interests are enriched and prepared for sequencing using the Agilent SureSelect system. Sequencing is performed using Illumina technology and reads are aligned to the reference sequence. Variants are identified and evaluated using a custom collection of bioinformatic tools and comprehensively interpreted by our team of directors and genetic counselors. All pathogenic and likely pathogenic variants are confirmed by Sanger sequencing. The technical sensitivity of this test is estimated to be >99% for single nucleotide changes and insertions and deletions of less than 20 bp.
Our CNV detection algorithm was developed and its performance determined for the sole purpose of identifying deletions and duplications within the coding region of the gene(s) tested. Partial exonic copy number changes and rearrangements of less than 400 bp may not be detected by this methodology. Regions of high homology and repetitive regions may not be analyzed. This methodology will not detect low level mosaicism, balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype. The sensitivity of our deletion/duplication assay may be reduced when DNA extracted by an outside laboratory is provided.

**Albinism Panel**

- **Sample specifications:** 3 to 10 cc of blood in a purple top (EDTA) tube
- **Cost:** $2500
- **CPT codes:** See test page
- **Turn-around time:** 8 weeks

*Note: We cannot bill insurance for the above test.*

**Results:**

Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

*For more information about our testing options, please visit our website at dnatesting.uchicago.edu or contact us at 773-834-0555.*

**References:**


*Committed to CUSTOMIZED DIAGNOSTICS, TRANSLATIONAL RESEARCH & YOUR PATIENTS’ NEEDS*