Leukodystrophy NGS Panel

Description
The Leukodystrophy NGS Panel analyzes 137 genes known to cause leukodystrophies and genetic leukoencephalopathies. The genes within this panel are recommended by the Global Leukodystrophy Initiative (GLIA) consensus for the diagnosis of these conditions. This approach to genetic testing can correctly identify the source of the patient's disorder quickly. This allows for improved patient management decisions, including avoiding unnecessary interventions and diagnostic testing, and identifying treatable conditions.

Although this NGS panel is comprehensive and consists of 137 genes, new genes are being identified as disease-causing for these conditions. This has lead to GLIA to also recommend Whole Exome Sequencing as a complementary test for when a definitive diagnosis is not arrived at with the Leukodystrophy NGS Panel.

The Leukodystrophy NGS Panel has sensitivity and specificity of >99% while the average depth is 124X. It covers all coding exons and 10 base pairs of flanking intronic sequences for all targeted genes. All reported variants are confirmed with Sanger sequencing to ensure a high level of analytical specificity. Parental testing is offered to further explore the pathogenicity of all variants of unknown significance (VUS) identified in the patient that may be sufficient to cause disease.

<table>
<thead>
<tr>
<th>CPT code</th>
<th>Please call our Reimbursement Specialists at 1-877-274-9432.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Test code</td>
<td>45000</td>
</tr>
<tr>
<td>Turnaround time</td>
<td>8 - 10 weeks</td>
</tr>
<tr>
<td>Platform</td>
<td>Illumina MiSeq platform</td>
</tr>
<tr>
<td>Sample types accepted</td>
<td>4 ml whole blood in a purple-top EDTA tube; ambient temperature</td>
</tr>
</tbody>
</table>
Disorders covered in this panel:
TPol-III related disorders, Hypomyelinating leukodystrophy, Dystonia type 4, HABC, Hypomyelination and congenital cataract, Pelizaeus-Merzbacher disease (PMD), Hypomyelinating leukodystrophy 2 (HLD2), Pelizaeus-Merzbacher-like disease (PMLD1), SOX10-associated PCWH, X-linked Adrenoleukodystrophy, Adult-onset leukodystrophy, Aicardi-Goutieres Syndrome (AGS), Alexander disease, Canavan disease, Adult-onset autosomal dominant leukodystrophy, Cerebrotendinous Xanthomatosis (CTX), Chloride Ion Channel 2 (CIC-2) related leukoencephalopathy with intramyelinic oedema, eIF2B related disorder, Fucosidosis, Globoid cell leukodystrophy (Krabbe disease), Hypomyelination with Brainstem and Spinal Cord involvement and lactate elevation (LBSL), Leukoencephalopathy with thalamus and brainstem involvement and high lactate (LTBL), Multiple sulfatase deficiency, Megalencephalic leukoencephalopathy with subcortical cysts (MLC), Metachromatic leukodystrophy (MLD), Oculodentodigital dysplasia (ODDD), Polyglucosan body disease (PGBD), RNAse T2 deficient leukoencephalopathy, Sialic acid storage disease, Sjögren-Larsson syndrome, Progressive leukoencephalopathy with ovarian failure, Peroxisome biogenesis disorder 1A,B, Peroxisome biogenesis disorder 5A,B, Peroxisome biogenesis disorder 10A, Peroxisome biogenesis disorder 4A,B, Peroxisome biogenesis disorder 3A,B, D-bifunctional protein deficiency, Sterol carrier protein 2 deficiency, Peroxisomal acyl-coA-oxidase deficiency, L2-Hydroxyglutaric aciduria, Cockayne syndrome, Deafness-dystonia-cerebral hypomyelination, Trichothiodystrophies, Hypomyelinating leukodystrophy 3, Hypomyelinating leukodystrophy 4, Global cerebral hypomyelination, GM1-gangliosidosis, GM2-ganliosidosis, Infantile neuronal ceroid lipofuscinosis, Mucolipidosis IV, Nasu-Hakola disease, Phosphoglycerate dehydrogenase deficiency, Allan-Herndon-Dudley syndrome, Band-like intracranial calcification with simplified gyration and polymicrogyria, Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL), Hereditary angiopathy with neuropathy-aneurysms-muscle cramps, Wilson disease, Menkes disease, Fabry disease, Familial hemophagocytic lymphohistiocytosis 5, Familial hemophagocytic lymphohistiocytosis 3, Familial hemophagocytic lymphohistiocytosis 4, Familial hemophagocytic lymphohistiocytosis 2, Lowe disease, Neimann-Pick type C2, Neimann-Pick type C1, Mitochondrial complex I disorders, Mitochondrial complex II disorders, Mitochondrial III disorders, Mitochondrial IV disorders, Mitochondrial complex V disorders, Glutaric Acidemia IIIC, Coenzyme Q10 deficiency (primary 1, 4, 5), Combined oxidative phosphorylation deficiency 1, Combined oxidative phosphorylation deficiency 2, Combined oxidative phosphorylation deficiency 4, Mitochondrial DNA depletion syndrome, Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome 1, Mitochondrial DNA depletion syndrome 5, Mitochondrial DNA depletion syndrome 7, Mitochondrial DNA depletion syndrome 8A, Mitochondrial DNA depletion syndrome 12, Spastic paraplegia 4 (SPG4), Spastic paraplegia 5 (SPG5), Spastic paraplegia 7 (SPG7), Spastic paraplegia 11 (SPG11), Spastic Paraplegia 13, Spastic paraplegia 15 (SPG15), Spastic paraplegia 20 (SPG20), Spastic paraplegia 21 (SPG21), Spastic paraplegia 35 (SPG35), Spastic paraplegia 56 (SPG56).
Genes Analyzed

AARS2, ARS2, ABCD1, ACOX1, ADAR, ADCK3, AIMP1, ALDH3A2, ATPAF2, ATP7A, ATP7B, ARSA, ASPA, BCAP31, BCS1L, BEST1, CLCN2, COL4A1, COQ2, COQ9, COX10, COX15, CSF1R, CYP2U1, CYP7B1, CYP27A1, C10orf2, DARS, DARS2, DGUOK, D2HGDH, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ERCC2, ERCC3, ERCC6, ERCC8, ETFDH, FA2H, FAM126A, FUCA1, GALC, GBE1, GFAP, GFM1, GJA1, GJC2, GLA, GLB1, GTF2H5, HEPACAM, HEXA, HSD17B4, HSPD1, HTRA1, L2HGDH, LMNB1, MCOLN1, MCL1, MPLKIP, MRPS16, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFAF1, NDUFV1, NOTCH3, NPC1, NPC2, OCLN, OCRL, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHGDH, PHYH, PLP1, POLG, POLG2, POLR3A, POLR3B, PPT1, PRF1, PSAP, PSAT1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RRM2B, SAMHD1, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SLC16A2, SLC17A5, SLC25A1, SLC25A4, SLC25A12, SOX10, SPAST, SPG7, SPG11, SPG20, SPG21STXB2, STX11, SUCLA2, SUMF1, SURF1, TACO1, TUFM, TREM2, TREX1, TYMP, TYROBP, UNC13D, ZFYVE26

We are committed to advancing your clinical practice

Transgenomic offers:

**Genetic testing panels**
We provide the most comprehensive genetic testing panels available for leukodystrophy and associated disorders.

**Professional support and resources**
Our scientific and genetic counseling team is here to support you with everything from test selection to outcomes research, analysis, and interpretation.

**Test reports**
Clear and comprehensive genetic testing reports assist you in making effective and timely patient management decisions.

To see our comprehensive test menu and request a visit from a Molecular Diagnostic Specialist, please visit [labs.transgenomic.com](http://labs.transgenomic.com) today or call us at 1-877-274-9432.