Noonan Syndrome/RASopathy Disorders NGS Panel

Description
The Noonan Syndrome/RASopathy next-generation sequencing (NGS) panel analyzes 11 genes associated with Noonan syndrome and/or other related disorders in the RASopathy pathway. RASopathy refers to a group of developmental syndromes caused by mutations in genes belonging to the RAS/MAPK pathway and includes disorders such as Noonan syndrome, Noonan-like syndrome, LEOPARD syndrome, Costello syndrome, autoimmune lymphoproliferative syndrome, and cardiofaciocutaneous syndrome. RASopathies are inherited in an autosomal-dominant manner and mutations are either de novo or, in some instances, inherited from a parent. Features seen in RASopathy disorders vary greatly, but some common features include short stature, congenital heart defects, developmental delays/intellectual disabilities, vision and hearing problems, broad or webbed neck, low muscle tone, skin problems, and characteristic facial features. The incidence of RASopathies range from about 1 in 1,000 in Noonan syndrome to very rare in cardiofaciocutaneous syndrome (only 300 individuals reported).

This NGS panel has sensitivity and specificity of >99% while the average read depth is 650X. It covers all coding exons and 10 base pairs of flanking intronic sequences for all targeted genes. High Resolution Deletion/Duplication testing is performed on the genes in this panel. This allows for enhanced detection abilities of both whole gene and intragenic dosage anomalies.

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.

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<tr>
<th>CPT code</th>
<th>Please call our Reimbursement Specialists at 1-877-274-9432.</th>
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<tr>
<td>Test code</td>
<td>40000</td>
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<td>Turnaround time</td>
<td>12 weeks</td>
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| Platform | • Illumina MiSeq and a laboratory-developed bioinformatics pipeline.  
• High Resolution Deletion/Duplication Analysis |
| Sample types accepted | 4 ml whole blood in a purple-top EDTA tube; ambient temperature |
**Genes Analyzed**
BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1

**References:**

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**.