About EGL Genetics

EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

Noonan Syndrome and Related Disorders

The Ras/mitogen-activated protein kinase (MAPK) pathway is a major pathway involved in the control of the cell cycle, differentiation, growth, and senescence. Any disruption in this pathway, due to germline mutations in the genes involved, results in a class of congenital developmental syndromes described collectively as the RASopathies.

RASopathies, while distinct syndromes, share some overlapping features such as craniofacial dysmorphology; varying degrees of neurocognitive impairments; cutaneous, ocular, and musculoskeletal abnormalities; and cardiac malformations. Some syndromes are also associated with an increased risk of cancer.

The Noonan Syndrome and Related Disorders Panel tests for 13 genes associated with the following conditions:

• Noonan syndrome
• LEOPARD syndrome
• Cardiofaciocutaneous (CFC) syndrome
• Costello syndrome
• Noonan-like syndrome with loose anagen hair

<table>
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<tr>
<th>Genes Included on Noonan Syndrome and Related Disorders Panel*</th>
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<tbody>
<tr>
<td>BRAF</td>
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<td>CBL</td>
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*Please note that deletion/duplication analysis is not completed for all genes in the panel. Some genes on this panel are associated with additional phenotypes.

All genes on the next generation sequencing panel may be ordered separately. Genes included on panels are subject to change. Please visit www.genetcslab.emory.edu/Noonan for current panel information.

Test Code | Test Name | CPT*** Codes
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MNOO1 | Noonan Syndrome and Related Disorders: Sequencing Panel | 81406 (x1)

***CPT* is a registered trademark of the American Medical Association.

References: