Prima Cancer Genetics

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Hemophagocytic Lymphohistiocytosis Gene Panel

About

Hemophagocytic lymphohistiocytosis (HLH) is a rare disease affecting predominantly children but also affects adults, characterized by hyperinflammatory features and life-threatening. A subset of patients, referred to as having familial hemophagocytic lymphohistiocytosis (FHL), have various underlying genetic abnormalities.

It is mainly characterized by proliferation and infiltration of hyperactivated macrophages and T-lymphocytes manifesting as acute illnes, prolonged fever, cytopenias, and hepatosplenomegaly. In case of familial hemophagocytic lymphohistiocytosis, the diagnosis is made on the presence of clinical presentation/criteria and is confirmed by molecular genetic testing.

Acquired or reactive secondary HLH is difficult to distinguish from familial/primary HLH clinically or by histologic findings alone. By the advancement in genetic diagnosis of FHL, molecular genetic testing is strongly recommended even in HLH suspected to be acquired. Moreover. HLH gene panel also helps in differential diagnosis

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HLH gene panel by MedGenome

- CAP accreditation for NGS panels, clinical exome, whole exome and other confirmatory testing
- Advanced sequencing technologies, automated scripts and precise bioinformatic pipelines and superior analytical performance
- Targeted sequencing represents a cost-effective approach to detect variants present in multiple/large genes in an individual
- Detects both SNPs and small InDels in the coding regions and splice site junctions
- Insilico analysis to detect CNVs (Copy Number Variations)
- Variants are strictly classified based on ACMG guidelines
- Systemic variant analysis and clinical interpretation workflow
- Reviewed by a team of skilled scientists and clinical geneticists

Genes Covered

AP3B1, BLOC1S6, CD27, ITK, LYST, MAGT1, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, XIAP

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