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TRANSFORMING COMPLEX DATA
INTO CLINICAL ANSWERS

LOSS OF CD55 IN ECULIZUMAB-RESPONSIVE PROTEIN-LOSING ENTEROPATHY

RAMBAM Health Care Campus

Applying Genoox's WES family analysis. The link between CD55 deficiency and the protein-losing enteropathy phenotype was not reported in the literature to date.

Protein-losing enteropathy usually manifests as peripheral and visceral edema due to hypoalbuminemia. The genetic lab at the Rambam Health Care Campus followed a large consanguineous Muslim–Arab family that included six patients who had protein-losing enteropathy associated with hypercoagulability. The laboratory was able to identify a homozygous frameshift variant in CD55 segregating with a hypercoagulability associated protein-losing enteropathy, applying Genoox's WES family analysis. The link between CD55 deficiency and the protein-losing enteropathy phenotype was not reported in the literature to date.

The Process

Whole-exome sequencing analysis was performed using Genoox's advanced family analysis which supports all known inheritance models. By combining proprietary machine learning algorithms, Genoox was able to promptly identify a rare homozygous frameshift variant in CD55 (ClinVar accession number, SCV000579315).

The variant was later validated by examining segregation with the disease phenotype in the extended family.

Congenital CD55 deficiency is very rare; to date, only nine patients with CD55 deficiency have been described. Four of them had gastrointestinal phenotypes, including protein-losing enteropathy, which thereby supports the link between CD55 deficiency and the protein-losing enteropathy phenotype.

The Result

Following these findings, the patients were treated with eculizumab, a terminal complement inhibitor, which has proven therapeutic benefits in the treatment of other disorders of complement dysregulation, such as CD59 deficiency patients presented with a decrease (by approximately 60%) in complement activation, and a steady increase in serum albumin and total protein concentrations, accompanied with a decrease in the number of stools and improvement in stool consistency.

Key Takeaways

Genoox has developed a number proprietary tools to help clinicians reach rapid clinical insights, such as a Superior Relevance Evidence Scoring (G-RES) engine. Based on artificial intelligence technology, G-RES is able to rapidly identify, prioritize, and classify the relevant, disease causing pathogenic variants from less significant variant mutations. Its extremely high variant-detection sensitivity provides an exceptional level of confidence with variant calls, as well as a significant reduction in sequencing costs and analysis man-hours.

Our Rapid processing engine (G-GQL®) offers built-in quality systems which ensure accurate and targeted patient care, and avoid the costly errors associated with missing an important correlated variant, or producing noisy results due to false positives calls.

Our exhaustive analysis utilizes all existing and breaching published research from both curated and uncurated databases, as well as individual journals and papers using direct matching, semantical associations and other investigative techniques. Our software updates ensure that both historical data, as well as the latest published research is included in your result.

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