Fanconi Anemia Report

Introduction

The Fanconi Anemia test is based on Whole Genome Sequencing Test. As such, it analyzes all Common and Rare Variants associated with Fanconi Anemia disease instead of a limited set of genes. Fanconi anemia is a rare autosomal recessive disorder characterized by several phenotypic abnormalities, also present with rare heterosome forms. The most important aspect of the condition is related to the function of the bone marrow, which fails to produce white blood cells, red blood cells or platelets. Congenital malformations have intrafamilial variability. When congenital malformations are not evident, the diagnosis can be delayed until the onset of bone marrow failure (BMF).

In our analysis, we found pathogenic or likely pathogenic variants related to:

- Unknown

Genes/Locations included in report:

ATM (0)	ATR (0)	BLM (1)	NBN (0)	SLX4 (0)	BRCA2 (1)	BRIP1 (1)
CXCR4 (0)	ERCC4 (0)	FANCA (1)	FANCB (0)	FANCC (0)	FANCE (0)	FANCF (1)
FANCG (0)	FANCI (0)	FANCL (0)	FANCM (0)	PALB2 (0)	XRCC2 (0)	FANCD2 (0)
RAD51C (0)						

Variants Found:

Gene/Loc	Chr: Pos	RSID	Phenotype Name	Zygosity	Variant	Allele Frequency	Significance	Review Status
BRCA2	chr13:32906729	rs1555281742	Unknown	HET	A>C		pathogenic	★
BLM Rare	chr15:91352442	rs760554566	Bloom syndrome	HET	C>A	0.00001	uncertain significance	★
FANCF	11:22644893	rs45554234	Fanconi anemia	HOM	A>AT		uncertain significance	★
BRIP1	17:59757841	rs1555571892	Neoplasm of the breast	HET	C>CTTTCTT		uncertain significance	★★
FANCA	16:89883148	rs11275235	not specified	HET	A>AGGCCTTGCGTCGT		uncertain significance	★

List of Conditions:

- Unknown

Methods

Extraction

Before sequencing, DNA extraction and library preparation processes were carried-out by automated liquid handling robots. Sequencing was completed using the NovaSeq 6000 instrument (Illumina).

The Nextera DNA Flex (Illumina) library was used during sequencing.

Analysis

Primary and secondary analysis was performed on the Illumina DRAGEN platform. Our secondary analysis extends the GATK 'best practices' pipeline. This includes Variant Quality Score Recalibration

It is important to note that applying a filter will not remove any data from the VCF file; it will just annotate the "FILTER" column. Variants with the "PASS" annotation are considered high quality and may, therefore, be used for advanced downstream analysis.

Sequence data is primarily aligned to the GATK GRCh37 reference genome and mitochondria is aligned to the Revised Cambridge Reference Sequence (NC_012920.1). Additional references may have been requested though tertiary analysis is not conducted on variant calls using references other than GRCh37.

Limitations

Test results are not interpretations. All variants reported in the genes included in the panel are reported.

Rare polymorphisms may lead to false-negative or false-positive results.

Due to limited read length and other contributing technical limitations, repeat expansions (e.g. in the Huntington gene, the SCA-genes, the myotonic dystrophy repeat region, and other similar regions) cannot be assessed with the applied method

Disclaimer

Any preparation and processing of a sample from saliva collection kit to Dante Labs by a customer is assumed to belong to the email used by the customer at the moment of kit registration on the Dante Labs Genome Manager platform before the shipment of the specimen to the laboratory.

The analysis and reporting conducted by Dante Labs are based on information from one or more published third-party scientific and medical studies.

Because of scientific and medical information changes over time, your risk assessment for one or more of the conditions contained within this report may also change over time. For example, opinions differ on the importance and relative weights given to genetic factors. Also, epidemiological data isn't available for some conditions, and this report may not be able to provide definitive information about the severity of a particular condition. We recommend asking your healthcare provider to correctly interpret them. Therefore, this report may not be 100% accurate (e.g., new research could mean different results) and may not predict actual results or outcomes.

This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained.

Contact

Please contact contact@dantelabs.com for more information on the contents of this report, our analysis methodology, and the limitations of this process.