Fanconi Anemia

Precision Panel

Overview

Fanconi Anemia (FA) is an inherited bone marrow failure syndrome featuring pancytopenia, predisposition to malignancy and characteristic physical abnormalities such as short stature, developmental delay, thumb abnormalities, among others. It is the most frequently reported of the rare inherited bone marrow failure syndromes. Individuals with FA require increased surveillance for malignancies and organ dysfunction. The defect relies in the inability to repair deleterious types of DNA damage resulting in genomic instability which in turn leads to increased risk of malignancy and defective haematopoiesis. In most cases, FA is inherited in an autosomal recessive manner although X-linked and autosomal dominant patterns exist.

The Igenomix Fanconi Anemia Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of pancytopenia ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Fanconi Anemia Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Short stature
- Skin pigmentation
- Petechiae
- Bruises
- Pallor
- Fatigue
- Frequent infections
- Thumb or other radial ray abnormalities
- Other malformations: congenital heart disease, gastrointestinal anomalies, CNS anomalies etc

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form hematopoietic stem cell transplantation as well as preventive measures such as active restriction, management of acute complications and early and continuous surveillance of malignancy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	HPO INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
BRCA1	Familial Breast-ovarian Cancer, Fanconi Anemia Complementation Group S, Familial Pancreatic Carcinoma, Primary Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Familial Breast-ovarian Cancer, Fanconi Anemia Complementation Group D1, Glioma Susceptibility, Medulloblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor, Nephroblastoma	AD,AR,MU	98.51	3343 of 3451
BRIP1	Fanconi Anemia Complementation Group J, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	94.97	235 of 237
ERCC4	Fanconi Anemia Complementation Group Q, Xeroderma Pigmentosum Complementation Group F, Cockayne Syndrome,	AR	99.68	69 of 72
FANCA	Fanconi Anemia	AR	95.17	497 of 502
FANCB	Fanconi Anemia Complementation Group B, Vacterl Association With Hydrocephalus	X,XR,G	95.53	NA of NA
FANCC	Fanconi Anemia Complementation Group C	AR	100	75 of 75
FANCD2	Fanconi Anemia Complementation Group D2	AR	100	62 of 63
FANCE	Fanconi Anemia Complementation Group E	AR	97	17 of 18
FANCF	Fanconi Anemia Complementation Group F	AR	99.31	17 of 18
FANCG	Fanconi Anemia Complementation Group G		100	94 of 94
FANCI	Fanconi Anemia Complementation Group I	AR	100	53 of 54
FANCL	Fanconi Anemia Complementation Group L	AR	100	25 of 26
FANCM	Premature Ovarian Failure, Spermatogenic Failure, Fanconi Anemia	AR	99.73	59 of 61
MAD2L2	Fanconi Anemia Complementation Group V	AR	99.91	1 of 1
PALB2	Breast Cancer, Fanconi Anemia Complementation Group N, Familial Pancreatic Carcinoma, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	98.78	601 of 617
RAD51	Fanconi Anemia Complementation Group R, Hereditary Breast And Ovarian Cancer Syndrome	AD	99.98	16 of 16
RAD51C	Fanconi Anemia Complementation Group O, Hereditary Breast And Ovarian Cancer Syndrome	AR	100	130 of 130
RFWD3	Fanconi Anemia Complementation Group W, Fanconi Anemia	AR	99.99	2 of 2
SLX4	Fanconi Anemia Complementation Group P	AR	99.92	76 of 76
UBE2T	Fanconi Anemia Complementation Group T	AR	100	4 of 4
XRCC2	Fanconi Anemia Complementation Group U	AR	98.39	28 of 28

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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