DISEASE:
Fanconi anemia

NAME:	Fanconi anemia
DESCRIPTION:	A rare genetic multisystem disorder characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors.
ORPHACODE:	84
SYNONYMS:	Fanconi pancytopenia

XREF(S):	Orphanet OMIM OMIM OMIM OMIM MeSH MedDRA ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	RAD51 MAD2L2 BRCA1 XRCC2 RFWD3 BRCA2 BRIP1 ERCC4 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCL FANCM FANCI PALB2 RAD51C SLX4 UBE2T
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RELATED CONTENT

Related Genetic Tests

- Fanconi anemia (FANCC) (hot spot mutation - c.456+4A>T)
- Fanconi anemia (gene panel)
- Hereditary cancer (Breast, ovary, colon) (26 genes)
- Hereditary cancer (gene panel)
- Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan) (4 genes; 7 hot spot mutations)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent

Related Analytes

- BRCA1 DNA repair associated
- BRCA2 DNA repair associated
- BRCA1 interacting helicase 1
- ERCC excision repair 4, endonuclease catalytic subunit
- FA complementation group A
- FA complementation group B
- FA complementation group C
- FA complementation group D2

- FA complementation group E
- FA complementation group F
- FA complementation group G
- FA complementation group I
- FA complementation group L
- FA complementation group M
- mitotic arrest deficient 2 like 2
- partner and localizer of BRCA2
- RAD51 recombinase
- RAD51 paralog C
- ring finger and WD repeat domain 3
- SLX4 structure-specific endonuclease subunit
- ubiquitin conjugating enzyme E2 T
- X-ray repair cross complementing 2

Related Gene Panels

- Hot spot mutation among Jewish (4 genes, 7 mutations) - UZA
- Cancer (Breast, ovary, colon,…) (26 genes) - ULG
- Fanconi anemia - UGent
- Hereditary predisposition to cancer (47 genes) - IPG
