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Review

Fanconi anemia

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Abstract

Fanconi anemia (FA) is a rare human recessive syndrome featuring bone marrow failure, myelodysplasia, and predisposition to cancer as well as chromosome fragility and hypersensitivity to DNA interstrands crosslinking agents. FA was described in 1927 by the Swiss pediatrician Giuseppe Fanconi, which reported a first family with three affected sibling presenting developmental defects and anemia.

FA cells are hypersensitive, at both cellular and chromosomal levels, to the exposure to DNA interstrands crosslinking agents, like mitomycin C, diepoxybutane, cis-platinum or photoactivated psoralen.

The chromosomal response to DNA interstrands crosslinks (ICLs)-inducing agents is so typical that the observation of both the induced frequency of chromosome aberrations and their type, i.e. tri- and quadri-radials, is considered the best diagnostic criteria for FA.

Indeed, looking simply at the clinical hallmarks of the patients, it is difficult to distinguish FA patients from several other bone marrow failure syndromes. Alternatively, since the FA cells need more time to pass through both G2 and S phases than normal cell, the analysis by flow cytometry of the over accumulation of the FA cells in G2 following exposure to ICL-inducing agents could be a useful approach for diagnosis.

More recently, molecular and biochemical approaches looking at gene mutations, proteins expression and/or post-translational modifications are used to validate cytogenetics conclusions.

To date 19 different genes (FANC) have been associated to FA.

The FANC proteins constitute a pathway which essential function is to deal with replication stress assuring the transmission of a stable genome from one cell to the daughters and acting both during replication, to cope with stalled replication forks, but also in G2 and M phases, to resolve un-replicated or not fully replicated regions before telophase.

For review: Duxin and Walter, 2015; Bogliolo and Surralles, 2015; Walden and Deans, 2014; Soulier 2011; Lobitz and Velleuer, 2006.

Keywords

Fanconi anemia, DNA repair, Replication, Acute Myeloid Leukemia, Bone Marrow Failure.

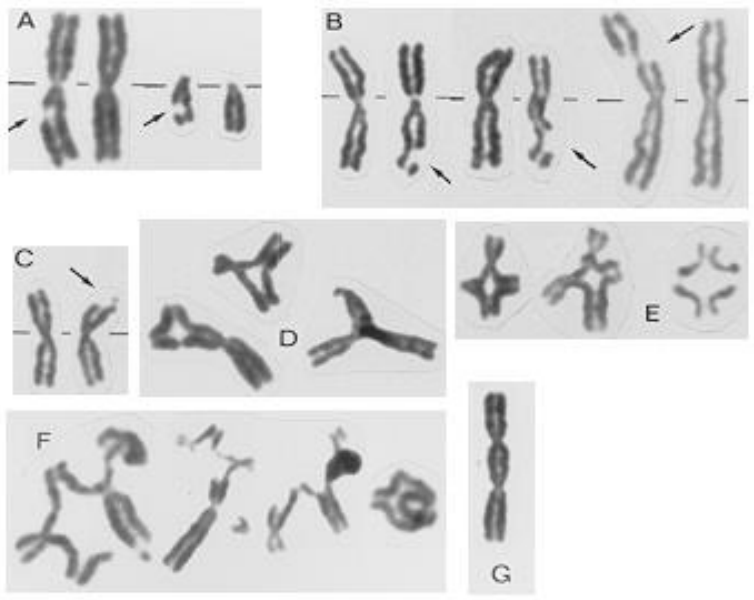
Identity

Other names

Fanconi pancytopenia

Note

Nineteen genes currently involved (for which bi-allelic inactivating mutations are retrieved in affected individual): FANCA, FANCB, FANCC, BRCA2 (FANCD1), FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, BRIP1 (BACH1/FANCJ) FANCL, FANCM, PALB2 (FANCN), RAD51C (FANCO), SLX4 (FANCP), ERCC4 (FANCQ/XPF) RAD51 (FANCR), BRCA1 (FANCS), UBE2T (FANCT).



A: gaps; B: breaks; C: deletion; D: triradials; E: quadriradials; F: complex figures; G: dicentric. Giemsa staining - Jean Loup Huret.

Inheritance
Autosomal recessive and X-linked (for FANCB); the estimated prevalence is 1 to 5 cases for million people; with a heterozygous carrier frequency of around 1/300 people.

Cytogenetics

Inborn conditions

Spontaneous elevated levels of chromatid and chromosome gaps and breaks, presence of abnormal figures, in particular triradials and quadriradials.
Hypersensitivity to the clastogenic effects of DNA crosslinking agents, like mitomycin C, diepoxybutane or cis-Platin.

Cytogenetics of cancer

Clonal abnormalities were reported in MDS and AML: in particular: -5/del(5q) and -7/del(7q) .

Other findings

slowing of the cell cycle (G2/M transition, with accumulating of cells in G2)
impaired oxygen metabolism
defective P53 induction

Genes involved and proteins

Note
To date 19 complementation groups have been described (A to T). FANCA represents 60 to 70% of the patients, FANCC and FANCG (10 to 15% each), meaning that all the other are extremely rare (less than 3% each).
In response to DNA damage and together with several other partners involved in DNA damage signaling and cell cycle checkpoint activation, the FANC proteins work long a linear pathway to cope with the replication stress induced by the presence of DNA lesions and help in the replication rescue by homologous recombination based mechanism.
Briefly, FANCA, FANCB, FANCC, FANCE, FANCF, FANCG and FANCL (with other companion proteins) assemble on FANCM and meet UBE2T to monoubiquitinate FANCD2 and FANCI.
Following their monoubiquitination, the FANCD2/FANCI heterodimer assembles into subnuclear foci where in a yet undetermined manner participates to and/or coordinates the elimination of the lesions and the restart of the stalled replication fork thanks to the action of the other component of the FANC pathway, which include structure specific endonucleases (XPF, SLX4) and homologous recombination proteins (RAD51, BRCA1, BRCA2, ...)
The pathway, or some of its components, participate also to transcription regulation, epigentics, production/response to inflammatory and stress induced cytokines and interferons.

FANCA (Fanconi anemia, complementation group A)

Location
16q24.3

Complementation Group	Extimated Frequency	Gene name	Additional Gene Name	Chromosomal Position	Protein M.W. (kDa)	Cloning Year
A	60-70	*FANCA*		16q24.3	160	1996
B	rare	*FANCB*	FAAP95	Xp22.2	95	2004
C	10-15	*FANCC*		9q22.3	60	1992
D1	1-5	*FANCD1*	BRCA2	13.q12.3	380	2002
D2	1-5	*FANCD2*		3p25.3	160	2001
E	rare	*FANCE*		6p21.3	60	2000
F	rare	*FANCF*		11p15	42	2000
G	10-15	*FANCG*	XRCC9	9p13.3	70	1998
I	rare	*FANCI*		15q26.1	150	2007
J	rare	*FANCJ*	BACH1; BRIP1	17q23.2	130	2005
L	rare	*FANCL*		2p16.1	52	2003
M	rare	*FANCM*		14q21.2	250	2005
N	rare	*FANCN*	PALB2	16p12.12	131	2007
O	rare	*FANCO*	RAD51C	17q22	42	2010
P	rare	*FANCP*	SLX4	16p13.3	200	2011
Q	rare	*FANCQ*	ERCC4; XPF	16p13.12	105	2013
R	rare	*FANCR*	RAD51	15q15.1	37	2015
S	rare	*FANCS*	BRCA1	17q21	220	2015
T	rare	*FANCT*	UBE2T	1q32.1	22	2015

Note

The gene spans 80kB and contains 43 exons. FANCA is the most frequently mutated among the 19 known FANC genes: it accounts for more than 60% of the FA patients worldwide.

Alternative splice results in the production of several transcripts variants encoding different protein isoforms.

The most representative protein is a polypeptide of 1455-amino acids weighting approximatively 163 kDa.

Present in both cytoplasms and nucleus, the protein possesses a nuclear localization signal but lacks of other known regulatory motifs and any biochemical function was ascribed to it.

FANCA participates to the nuclear FANCcore complex that hosts the E3 ligase (FANCL) activity that, in collaboration with the E2 UBE2T, monoubiquitinates FANCD2 and FANCI in response to DNA damage. FANCA interacts directly with FANCG and FAAP20.

Mutations

FANCB (Fanconi anemia complementation group B)

Location

Xp22.2

Note

FANCB is constituted by 10 exons spanning 77kB. Alternative splicing results in two transcript variants encoding a same protein of 859-amino acids with a MW of 98 kDa. Any biochemical function was reported for the protein.

FANCB aggregates with FANCL and FAAP100 in a sub-complex that participates to the FANCcore complex to mediate FANCD2 and FANCI monoubiquitination in response to DNA damage. FANCB stabilizes FANCL and needs FANCA to translocate into thAe nucleus. Mutations in FANCB are associated to both Fanconi anemia and X linked VACTERL with hydrocephalus syndromes.

FANCC (Fanconi anaemia complementation group C)

Location

9q22.32

Note

FANCC has been the first FANC gene to be cloned. It contains 14 exons and codes an ORF of 1677 bp which translation results in a protein of 558aa, weighting about 63kDa. The protein, present in both cytoplasm and nucleus, interacts with FANCE and FANCF, a subgroup participating to the FANCcore complex. Any direct biochemical function was reported for FANCC.

BRCA2 (breast cancer 2, early onset)

Location

13q13.1

Note

The gene contains 27 exons, coding a mRNA which translation results in a protein of 3418aa, weighting about 385kDa. The protein is involved in the homologous recombination process. FANCD1/BRCA2 contains several repetitions of a 70 aa motif called the BRC motif that mediate

RAD51 interaction. Indeed, FANCD1/BRCA2 is the cargo that target RAD51 to ssDNA stretches covered by RPA at DBS. It interacts with several proteins involved in DNA metabolism, including FANCD2, FANCN/PALB2, POLH and some components of the TREX-2 complex. FANCD1/BRCA2 inherited mutations are associated to the recessive syndrome Fanconi
anemia while carriers of one inactivated allele are at risk for breast and ovarian cancer predisposition following the somatic loss-of-function of the wild-type allele.

FANCD2 (Fanconi anemia, complementation group D2)

Location
3p25.3

Note
The gene contains 44 exons. FANCD2 encodes a 1,451-amino acid nuclear protein. As several other FANC proteins, FANCD2 had no known functional domains. With its major partner, FANCI, FANCD2 is the target of the Ubiquitin-ligase activity of the the FANCcore complex. In presence of DNA damage or replication stress, FANCD2 is monoubiquitinated on K561 and targeted to subnuclear foci where it colocalize with several DNA repair proteins. It is phosphorylated by both ATM and ATR. The protein participate to both replication safeguard and chromosome fragile sites integrity maintenance. Interacts directly or indirectly with several proteins, including, FANCI, FANCE. USP1, MEN1, BRCA1, BRCA2, phosphorylated FANCG, FAN1 and DCLRE1B/Apollo.

FANCE (Fanconi anemia, complementation group E)

Location
6p21.31

Note
The gene contains 10 exons. FANCE protein is constituted by 536 amino-acids weighting approximatively 59kDa. It contains two Nuclear Localization Signal (NLS). FANCE forms with FANCC and FANCF a FANCcore complex sub-complex. It is required for FANCC nuclear accumulation and connects the FANCcore complex to FANCD2 allowing the FANCL/UBE2T-mediated FANCD2 monoubiquitination. It is phosphorylated by CHK1 in response to DNA damage. As several other FANC proteins, FANCE had no known biochemical functions.

FANCF (Fanconi anemia, complementation group F)

Location
11p14.3

Note
FANCF is an intron-less gene. The protein, long of 374aa, weights 42kDa. FANCF is predominantly nuclear, where it interacts with FANCE and FANCC, a subgroup participating to the FANCcore complex. As a FANCcore complex participant, FANCF is involved in FANCD2 and FANCI monoubiquitination. FANCF had no known biochemical functions.

FANCG (Fanconi anemia, complementation group G)

Location
9p13.3

Note
The gene codes at least two mRNA of 2.2 and 2.5 kb, which translation results in a major proteins of 622 aa, weighting 68kDa.
It participates to the FANCcore complex and its phosphorylation on serine 7 is mandatory for its function inside the complex.
Nevertheless, as for several other FANC proteins any biochemical function has been attributed to FANCG. As for the other components of the FANCcore complex, its presence inside the complex is mandatory for FANCD2 and FANCI monoubiquitination and targeting to subnuclear foci.

FANCI (Fanconi anemia complementation group I)

Location
15q26.1

Note
The gene contains 38 exons. The FANCI protein is long of 1328aa, weights 50kDa and contains 3 NLS. FANCI is phosphorylated by ATM/ATR and is monoubiquitinated by the FANCcore complex on the lys523. It is considered as a functional homolog of FANCD2. The two proteins forms a heterodimer that, following their DNA damage- or replication stress -induced monoubiquitination, relocalizes to subnuclear foci to optimally restore DNA and rescue replication in a yet undetermined manner.

BACH1 (BTB domain and CNC homolog 1)

Location
21q21.3

Note
The gene encodes a protein of 1249aa with a molecular mass de 141kDa. Memeber of the RecQ DEAH helicase family, FANCJ interact with BRCA1 participating to the DNA double-strand breaks repair by homologous recombination. Germline mutations in FANCJ are associated to breast and ovarian cancer susceptibility. Biallelic inheritance results in a Fanconi anemia-like phenotype.

BRIP1 (BRCA1 interacting protein C-terminal helicase 1)

Location
17q23.2

Note
The gene encodes a protein of 1249aa with a molecular mass de 141kDa. Member of the RecQ DEAH helicase family, FANCJ interact with BRCA1 participating to the DNA double-strand breaks repair by homologous recombination. Germline mutations in FANCJ are associated to breast and ovarian cancer susceptibility. Biallelic inheritance results in a Fanconi anemia-like phenotype

FANCL (Fanconi anemia complementation group L)

Location
2p16.1

Note
It codes a proteins of 373 aa, weighting 43 kDa, containing 3 putative WD40 motifs and a PHD zync finger motif. The protein could be retrieved in both cytoplasm and nucleus. FANCL is the catalytic subunit of the FANCore complex. It has the E3 ubiquitin ligase activity necessary for FANCD2 and FANCI monoubiquitination. It mediate ubiquitin release from UBE2T and UBE2W.

FANCM (Fanconi anemia complementation group M)

Location
14q21.2

Note
It code for a protein of 2048aa. Contains an N-terminal helicase domain ans possess the ability to translocate on duplex DNA. It belongs to the DEAD box helicase family. It is hyperphosphorylated by ATR in response of DNA damage FANCM is thought be the transporter of the FANCcore complex along the DNA and, so, it participates de facto to both FANCD2 and FANCI optimal monoubiquitination.

PALB2 (partner and localizer of BRCA2)

Location
16p12.2

Note
FANCN contains 13 exons and encodes for a protein of 1186 aa having a molecular mass of about 130kDa. The protein participates to homologous recombination in collaboration with its major partner BRCA2. It interacts also with BRCA1, RAD51, RAD51C and POLH. Monoallelic PALB2 mutations confer predisposition to breast and pancreatic cancers. hereditary bi-allelic mutations in FANCN result in Fanconi anemia.

RAD51C (RAD51 paralog C)

Location
17q22

Note
Member of the RAD51 gene family, involved in homologous recombination repair of damaged DNA and in meiotic recombination. RAD51C encodes a major 1.3 kB mRNA translated in a protein of 376 aa, weighting approximatively 45kDa. It interact with several DNA repair proteins, including RAD51 and PALB2.
It participates to several complexes with RAD51B, RAD51D and XRCC2 or with XRCC3. The monoallelic inheritance of RAD51C is associated to breast and ovarian cancers predispostion. The biallelic, recessive, inheritance of RAD51C mutations result in a Fanconi anemia-like syndrome.

SLX4 (SLX4 structure-specific endonuclease subunit)

Location
16p13.3

Note
The protein is constituted by 1834 aa which weights about 200kDa. Component of the SLX1-SLX4 structure-specific endonuclease, it is the docking platform of a complex assembling two other structure specific enducleases: XPF-ERCC1 and MUS81-EME1.
SLX4 is also associated to MSH2/MSH3, the telomere binding complex TRF2-RAP1 and the kinase PLK1. FANCP is required DNA repair, chromosome fragile sites maintenance and for replication fork failure rescue.

ERCC4 (xeroderma pigmentosum, complementation group F)

Location
16p13.12

Note
The gene contains 11 exons spanning more than 28 kb. The gene encodes 3 mRNA of 2.4, 3.8 and 7 kb, which translation results in a protein of 905 aa, having a mass of about 110 kDa. The protein interacts primarily with with ERCC1 making up the ERCC1-XPF-5' structure specific endonuclease. The protein also interacst with FANCP/SLX4. Biallelic inactivating mutation in this gene have been associated to Fanconi anemia, xeroderma pigmentosum, cockayne syndrome and XFE progeroid syndrome.

RAD51 (RAD51 recombinase)

Location

15q15.1

Note

Belonging to the RAD51 family, this gene is encodes several transcript variant, the major being a 1.8kb mRNA which translation results in a protein of 339aa weighting 37kDa which plays a central role in homologous recobination repair and in meiotic recombination. It interacts with BRCA1, BRCA2, RPA, and several other DNA repair proteins. The only Fanconi anemia patient associated to RAD51 mutation bears a de novo mutation which created a dominant-negative variant. Mutations in RAD51 have been also associated to breast cancer suceptibility and to the congenital Mirror Movements 2 syndrome.

BRCA1 (breast cancer 1, early onset)

Location

17q21.31

UBE2T (ubiquitin conjugating enzyme E2 T)

Location

1q32.1

Note

The gene contains 7 exons. Two transcript variants encodes different protein isoforms, the major is a protein of 197 amino acids weighting approximatively 22kDa. UBE2T is an E2-conjugating enzyme that collaborates with FANCL, the E3 ubiquitin ligase hosted by the FANC core complex, for the monoubiquitination of FANCD2 and FANCI. It interact with FANCL and BRCA1.

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