Rx-Genes
Treatment Guidance at the Point of Care

Disease

Fanconi anemia, complementation group J[1, 2]

Gene

BRIP1

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	guideline[3]
Non-Molecular Testing Availability	yes
Non-Molecular Test	MMC- and/or DEB-induced chromosome breakage analysis[4]
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1008
Disease Frequency (per 100,000)	0.76[5]
Genes Included in Disease Frequency	BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, ERCC4, FANCL, MAD2L2, RFWD3, SLX4, UBE2T
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Fanconi+Anemia&term=&cntry=&state=&city=&dist=

References

1 https://www.ncbi.nlm.nih.gov/books/NBK1401/

2 https://www.ncbi.nlm.nih.gov/pubmed/27869810

3 https://www.fanconi.org/images/uploads/other/Guidelines_4th_Edition.pdf

4 https://www.fanconi.org/images/uploads/other/Chapter_2_Guidelines_4th_Edition.pdf

5 https://www.ncbi.nlm.nih.gov/pubmed/21739583