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Full name:	Hematologic Familiar Forms - ULG
Laboratory:	Centre de Génétique Humaine - CHU Sart-Tilman
Created:	21 Dec 2021 - 21:51
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Related Diseases

- Acquired idiopathic sideroblastic anemia
- Acute myeloid leukaemia with myelodysplasia-related features
- Aggressive systemic mastocytosis
- Atypical chronic myeloid leukemia
- Autosomal dominant aplasia and myelodysplasia
- Autosomal dominant severe congenital neutropenia
- Autosomal recessive severe congenital neutropenia due to CSF3R deficiency
- Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency
- Autosomal thrombocytopenia with normal platelets
- Chronic neutrophilic leukemia
- Cyclic neutropenia
- Essential thrombocythemia
- Familial thrombocytosis
- Hereditary neutrophilia
- Idiopathic aplastic anemia
- Inherited acute myeloid leukemia
- Kostmann syndrome
- MIRAGE syndrome
- Pancytopenia-developmental delay syndrome
- Polycythemia vera
- Primary myelofibrosis
- RAS-associated autoimmune leukoproliferative disease
- Refractory anemia
- Refractory anemia with excess blasts type 1

- Refractory anemia with excess blasts type 2
- Shwachman-Diamond syndrome
- Wiskott-Aldrich syndrome
- X-linked severe congenital neutropenia

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ANKRD26	100.00	0	Genomic start 27389256 to genomic end 27389427 UTR position
ATG2B	99.33	0	No
ATM	98.90	0	No
ATR	99.19	0	No
ATRX	99.85	0	No
BLM	98.77	0	No
BRCA1	99.80	0	No
BRCA2	99.11	0	No
BRIP1	99.61	0	No
CBL	99.87	0	No
CEBPA	99.48	0	No
CHEK2	80.48	0	No
CSF3R	100.00	0	No
CTC1	99.99	0	No
DDX41	100.00	0	No

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
DKC1	100.00	0	Genomic start 153991031 to genomic end 153991240 UTR position
DNAJC21	98.55	0	No
EGLN1	98.71	0	No
ELANE	100.00	0	No
EPAS1	99.62	0	No
EPO	99.99	0	No
EPOR	100.00	0	No
ERCC4	98.33	0	No
ERCC6L2	99.48	0	No
ETV6	100.00	0	No
FANCA	99.93	0	No
FANCB	99.61	0	No
FANCC	99.72	0	No
FANCD2	99.00	0	No
FANCE	97.83	0	No
FANCF	100.00	0	No
FANCG	100.00	0	No
FANCI	99.66	0	No
FANCL	98.99	0	No

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
FANCM	97.86	0	No
G6PC3	100.00	0	No
GATA2	100.00	0	No
GFI1	100.00	0	No
GSKIP	99.99	0	No
HAX1	99.98	0	No
JAK2	97.81	0	No
KRAS	96.88	0	No
LIG4	99.98	0	No
MAD2L2	100.00	0	No
MECOM	99.83	0	No
MLH1	99.86	0	No
MPL	99.99	0	No
MSH2	98.36	0	No
MSH6	99.84	0	No
NBN	99.57	0	No
NF1	96.15	0	No
NHP2	99.79	0	No
NOP10	99.99	0	No

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
PALB2	99.80	0	No
PARN	99.84	0	No
PAX5	99.72	0	No
PMS2	98.87	0	No
PTPN11	95.76	0	No
RAD51	99.94	0	No
RAD51C	99.83	0	No
RBM8A	99.34	0	No
RPL11	99.02	0	No
RPL35A	98.16	0	No
RPL5	96.83	0	No
RPS10	98.48	0	No
RPS19	100.00	0	No
RPS24	98.39	0	No
RPS26	98.64	0	No
RPS7	88.11	0	No
RTEL1	100.00	0	Genomic start 62326911 to genomic end 62326911 and genomic start 62326900 to genomic end 62326928 and genomic start 62326958 to genomic end 62326986 intronic positions and 99.98 for CDS

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
RUNX1	99.94	0	No
SAMD9	100.00	0	No
SAMD9L	99.96	0	No
SBDS	96.51	0	No
SBF2	99.19	0	No
SHQ1	97.53	0	No
SLX4	99.98	0	No
SRP54	99.44	0	No
SRP72	97.98	0	No
STN1	99.75	0	No
TERC	100.00	0	Genomic start 169482849 to genomic end 169483098 UTR position
TERT	100.00	0	Genomic start 1295105 to genomic end 1295162 UTR position
TET2	99.97	0	No
THPO	100.00	0	No
TINF2	100.00	0	No
TP53	100.00	0	No
TPP1	99.99	0	No
UBE2T	98.01	0	No
USB1	100.00	0	No

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
VHL	99.98	0	No
VPS45	99.60	0	No
WAS	99.90	0	No
WRAP53	100.00	0	No
XRCC2	99.97	0	No