GENETIC TEST:
« Inherited bone marrow failures syndromes » with or without organ dysfunction

FULL NAME:	« Inherited bone marrow failures syndromes » with or without organ dysfunction
DESCRIPTION:	-Dedicated clinical form and patient's consent form are needed prior to the analysis (see attached form). -Check details on the CHU Liege's analytical web site: https://www.chu.ulg.ac.be/jcms/c2_25132963/fr/panel-maladies-hematologiques-hereditaires-whole-exome-sequencing
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA, Skin biopsy

METHOD CATEGORY:	Sequence analysis: entire coding region
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565552-565563
TURNAROUND TIME (MAXIMUM):	6 weeks
CREATED:	21 Dec 2021 - 21:29
CHANGED:	18 Mar 2024 - 10:00

RELATED CONTENT

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 Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

Related Analytes

- ankyrin repeat domain containing 26
- autophagy related 2B
- ATM serine/threonine kinase
- ATR serine/threonine kinase
- ATRX chromatin remodeler
- BLM RecQ like helicase
- BRCA1 DNA repair associated
- BRCA2 DNA repair associated
- BRCA1 interacting helicase 1
- Cbl proto-oncogene
- CCAAT enhancer binding protein alpha
- checkpoint kinase 2
- colony stimulating factor 3 receptor
- CST telomere replication complex component 1
- DEAD-box helicase 41
- dyskerin pseudouridine synthase 1
- DnaJ heat shock protein family (Hsp40) member C21
- egl-9 family hypoxia inducible factor 1
- elastase, neutrophil expressed
- endothelial PAS domain protein 1
- erythropoietin

- erythropoietin receptor
- ERCC excision repair 4, endonuclease catalytic subunit
- ERCC excision repair 6 like 2
- ETS variant transcription factor 6
- 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
- glucose-6-phosphatase catalytic subunit 3
- GATA binding protein 2
- growth factor independent 1 transcriptional repressor
- GSK3B interacting protein
- HCLS1 associated protein X-1
- Janus kinase 2
- KRAS proto-oncogene, GTPase
- DNA ligase 4
- mitotic arrest deficient 2 like 2
- MDS1 and EVI1 complex locus
- mutL homolog 1
- MPL proto-oncogene, thrombopoietin receptor
- mutS homolog 2
- mutS homolog 6
- nibrin
- neurofibromin 1
- NHP2 ribonucleoprotein
- NOP10 ribonucleoprotein
- partner and localizer of BRCA2

- poly(A)-specific ribonuclease
- paired box 5
- PMS1 homolog 2, mismatch repair system component
- protein tyrosine phosphatase non-receptor type 11
- RAD51 recombinase
- RAD51 paralog C
- RNA binding motif protein 8A
- ribosomal protein L11
- ribosomal protein L35a
- ribosomal protein L5
- ribosomal protein S10
- ribosomal protein S19
- ribosomal protein S24
- ribosomal protein S26
- ribosomal protein S7
- regulator of telomere elongation helicase 1
- RUNX family transcription factor 1
- sterile alpha motif domain containing 9
- sterile alpha motif domain containing 9 like
- SBDS ribosome maturation factor
- SET binding factor 2
- SHQ1, H/ACA ribonucleoprotein assembly factor
- SLX4 structure-specific endonuclease subunit
- signal recognition particle 54
- signal recognition particle 72
- STN1 subunit of CST complex
- telomerase RNA component
- telomerase reverse transcriptase
- tet methylcytosine dioxygenase 2
- thrombopoietin
- TERF1 interacting nuclear factor 2
- tumor protein p53
- tripeptidyl peptidase 1

- ubiquitin conjugating enzyme E2 T
- U6 snRNA biogenesis phosphodiesterase 1
- von Hippel-Lindau tumor suppressor
- vacuolar protein sorting 45 homolog
- WASP actin nucleation promoting factor
- WD repeat containing antisense to TP53
- X-ray repair cross complementing 2

Related Gene Panels

- Hematologic Familiar Forms - ULG
