GENETIC TEST:
Constitutional Mismatch Repair Deficiency Syndrome + Bloom syndrome (5 genes)

FULL NAME:	Constitutional Mismatch Repair Deficiency Syndrome + Bloom syndrome (5 genes)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis, Predictive and Pre-symptomatic diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565552-565563
ACCREDITATION (ISO 15189):	2021-07-08 / 2026-02-02
EQA:	• Lynch Syndrome
TURNAROUND TIME (MAXIMUM):	4 - 6 months

CREATED:	18 Jul 2019 - 13:15
CHANGED:	01 Mar 2023 - 14:55
URL:	https://laboboeken.nexuzhealth.com/pboek/internet/GHB%20CME/13380

Source URL: http://gentest.healthdata.be/genetic_test/72

RELATED CONTENT

Related Diseases

- Bloom syndrome
- Constitutional mismatch repair deficiency syndrome
- Lynch syndrome

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- BLM RecQ like helicase
- mutL homolog 1
- mutS homolog 2
- mutS homolog 6
- PMS1 homolog 2, mismatch repair system component

Related Gene Panels

- Constitutional Mismatch Repair Deficiency Syndrome / Bloom syndrome - KUL