GENETIC TEST:
X-linked hydrocephalia / CRASH (corpus callosum hypoplasia, retardation, adducted thumbs, spastic paraplegia, and hydrocephalus) syndrome (L1CAM gene)

FULL NAME:	X-linked hydrocephalia / CRASH (corpus callosum hypoplasia, retardation, adducted thumbs, spastic paraplegia, and hydrocephalus) syndrome (L1CAM gene)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Amniotic fluid, Chorionic villi, Cell culture
METHOD CATEGORY:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis

RIZIV CODE:	565471-565482
ACCREDITATION (ISO 15189):	2021-10-07 / 2026-06-14
EQA:	• DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger , • DNA Sequencing - Sanger
TURNAROUND TIME (MAXIMUM):	6 months (10 working days for prenatal diagnosis)
CREATED:	28 Aug 2019 - 14:45
CHANGED:	09 Mar 2023 - 16:22
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:X-gebonden%20hydrocefalie&&&&…

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