

GENETIC TEST:
Oculopharyngeal muscular dystrophy - PABPN1 gene GCN trinucleotide repeats

FULL NAME:	Oculopharyngeal muscular dystrophy - PABPN1 gene GCN trinucleotide repeats
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:	Targeted variant analysis
METHOD TECHNIQUE:	PCR based technique
RIZIV CODE:	565456-565460
TURNAROUND TIME (MAXIMUM):	3 months (10 working days for prenatal diagnosis)

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URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Oculopharyngale%20musculaire%...

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