GENETIC TEST:
Short stature/ Growth retardation/ (gene panel)

FULL NAME:	Short stature/ Growth retardation/ (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2023-11-09 / 2024-05-08
EQA:	• Familial SHOX-related disorders, • Familial SHOX-related disorders
TURNAROUND TIME (MAXIMUM):	6 months

CREATED:	21 Aug 2019 - 10:43
CHANGED:	22 Jan 2024 - 14:24
URL:	https://labogidsmedgen.uza.be/analyses/whole-exome-sequencing-wes-ikv-short-sta…

RELATED CONTENT

Related Diseases

- 3M syndrome
- Aarskog-Scott syndrome
- Achondroplasia
- Acromesomelic dysplasia, Maroteaux type
- Autosomal recessive Robinow syndrome
- Autosomal recessive primary microcephaly
- Coffin-Siris syndrome
- Cornelia de Lange syndrome
- DNA2-related mitochondrial DNA deletion syndrome
- Ear-patella-short stature syndrome
- Familial osteochondritis dissecans
- Floating-Harbor syndrome
- Growth delay due to insulin-like growth factor I resistance
- Growth delay due to insulin-like growth factor type 1 deficiency
- Hypochondroplasia
- Isolated growth hormone deficiency type IB
- Langer mesomelic dysplasia
- Laron syndrome
- Laron syndrome with immunodeficiency
- Léri-Weill dyschondrosteosis
- Melnick-Needles syndrome
- Microcephalic osteodysplastic primordial dwarfism type II
- Microcephalic primordial dwarfism, Dauber type
- Muenke syndrome
- Mulibrey nanism
- Otopalatodigital syndrome type 1

- Otopalatodigital syndrome type 2
- Peripheral resistance to thyroid hormones
- SHOX-related short stature
- Saethre-Chotzen syndrome
- Seckel syndrome
- Short stature due to growth hormone qualitative anomaly
- Short stature due to partial GHR deficiency
- Short stature-advanced bone age-early-onset osteoarthritis syndrome
- Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome
- Spondyloepimetaphyseal dysplasia, aggrecan type
- Spondyloepiphyseal dysplasia, Kimberley type
- Tall stature-long halluces-multiple extra-epiphyses syndrome
- Thanatophoric dysplasia type 1
- Thanatophoric dysplasia type 2
- Wiedemann-Steiner syndrome
- X-linked Ehlers-Danlos syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- aggrecan
- alkaline phosphatase, biomineralization associated
- ankyrin repeat domain containing 11
- archain 1
- AT-rich interaction domain 1A
- AT-rich interaction domain 1B
- AT-rich interaction domain 2

- bone morphogenetic protein 2
- B-Raf proto-oncogene, serine/threonine kinase
- Bruton tyrosine kinase
- Cbl proto-oncogene
- coiled-coil domain containing 8
- cell division cycle 45
- cell division cycle 6
- cyclin dependent kinase inhibitor 1C
- chromatin licensing and DNA replication factor 1
- CREB binding protein
- cullin 7
- 7-dehydrocholesterol reductase
- dishevelled segment polarity protein 1
- E1A binding protein p300
- FYVE, RhoGEF and PH domain containing 1
- fibroblast growth factor receptor 3
- filamin A
- growth hormone 1
- growth hormone receptor
- growth hormone releasing hormone receptor
- growth hormone secretagogue receptor
- geminin DNA replication inhibitor
- HESX homeobox 1
- high mobility group AT-hook 2
- HRas proto-oncogene, GTPase
- insulin like growth factor 1
- insulin like growth factor 1 receptor
- insulin like growth factor 2
- insulin like growth factor binding protein acid labile subunit
- Indian hedgehog signaling molecule
- lysine demethylase 6A
- lysine methyltransferase 2A
- lysine methyltransferase 2D

- KRAS proto-oncogene, GTPase
- leucine zipper like post translational regulator 1
- mitogen-activated protein kinase kinase 1
- mitogen-activated protein kinase kinase 2
- mitogen-activated protein kinase 1
- muscle RAS oncogene homolog
- NBAS subunit of NRZ tethering complex
- natriuretic peptide receptor 2
- NRAS proto-oncogene, GTPase
- obscurin like cytoskeletal adaptor 1
- origin recognition complex subunit 1
- origin recognition complex subunit 4
- origin recognition complex subunit 6
- pappalysin 2
- phosphoinositide-3-kinase regulatory subunit 1
- PLAG1 zinc finger
- POC1 centriolar protein A
- POU class 1 homeobox 1
- protein phosphatase 1 catalytic subunit beta
- protein tyrosine phosphatase non-receptor type 11
- Raf-1 proto-oncogene, serine/threonine kinase
- RAS p21 protein activator 2
- Ras like without CAAX 1
- RNA binding region (RNP1, RRM) containing 3
- receptor tyrosine kinase like orphan receptor 2
- RAS related 2
- SHOC2 leucine rich repeat scaffold protein
- SHOX homeobox
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1
- SOS Ras/Rac guanine nucleotide exchange factor 1
- SOS Ras/Rho guanine nucleotide exchange factor 2
- SRY-box transcription factor 3

- sprouty related EVH1 domain containing 2
- Snf2 related CREBBP activator protein
- signal transducer and activator of transcription 5B
- DNA topoisomerase III alpha
- tripartite motif containing 37
- Wnt family member 5A
- YY1 transcription factor

Related Gene Panels

- Growth retardation/short stature (genepanel) - UZA
