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Full name:	Short Stature (46 genes) - IPG
Version number:	Version 3
Laboratory:	Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
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Related Diseases

- 16q24.3 microdeletion syndrome
- 3M syndrome
- Aarskog-Scott syndrome
- Achondroplasia
- Acromesomelic dysplasia, Maroteaux type
- Autosomal dominant Robinow syndrome
- Autosomal recessive Robinow syndrome
- Camptodactyly-tall stature-scoliosis-hearing loss syndrome
- Coffin-Lowry syndrome
- Combined pituitary hormone deficiencies, genetic forms
- Costello syndrome
- Crouzon syndrome-acanthosis nigricans 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
- Hypothyroidism due to deficient transcription factors involved in pituitary development or function
- Isolated growth hormone deficiency type IB
- Isolated growth hormone deficiency type II
- KBG syndrome
- Kabuki syndrome
- Langer mesomelic dysplasia

- Laron syndrome
- Laron syndrome with immunodeficiency
- Léri-Weill dyschondrosteosis
- Muenke syndrome
- Mulibrey nanism
- Noonan syndrome
- Noonan syndrome with multiple lentigines
- Noonan syndrome-like disorder with juvenile myelomonocytic leukemia
- Noonan syndrome-like disorder with loose anagen hair
- Rubinstein-Taybi syndrome due to 16p13.3 microdeletion
- Rubinstein-Taybi syndrome due to CREBBP mutations
- Rubinstein-Taybi syndrome due to EP300 haploinsufficiency
- SHORT syndrome
- SHOX-related short stature
- Saethre-Chotzen syndrome
- Severe achondroplasia-developmental delay-acanthosis nigricans syndrome
- Short stature due to GHSR deficiency
- Short stature due to growth hormone qualitative anomaly
- Short stature due to isolated growth hormone deficiency with X-linked hypogammaglobulinemia
- Short stature due to partial GHR deficiency
- Short stature due to primary acid-labile subunit deficiency
- Short stature-advanced bone age-early-onset osteoarthritis syndrome
- Silver-Russell syndrome due to a point mutation
- Smith-Lemli-Opitz syndrome
- Spondyloepimetaphyseal dysplasia, aggrecan type
- Spondyloepiphyseal dysplasia, Kimberley type
- Symptomatic form of Coffin-Lowry syndrome in female carriers
- Tall stature-long halluces-multiple extra-epiphyses syndrome
- Thanatophoric dysplasia type 1
- Thanatophoric dysplasia type 2

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ACAN	90.00	1	NM_001369268.1

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ANKRD11	100.00	1	NM_013275.6
BRAF	100.00	1	NM_001354609.2
BTK	100.00	1	NM_000061.3
CBL	100.00	1	NM_005188.4
CCDC8	100.00	1	NM_032040.5
CREBBP	100.00	1	NM_004380.3
CUL7	100.00	1	NM_014780.5
DHCR7	100.00	1	NM_001360.3
DVL1	100.00	1	NM_001330311.2
EP300	100.00	1	NM_001429.4
FGD1	100.00	1	NM_004463.3
FGFR3	100.00	1	NM_001163213.1
GH1	100.00	1	NM_000515.5
GHR	100.00	1	NM_000163.5
GHRHR	100.00	1	NM_000823.4
GHSR	100.00	1	NM_198407.2
HRAS	100.00	1	NM_005343.4
IGF1	100.00	1	NM_000618.5
IGF1R	100.00	1	NM_000875.5
IGF2	100.00	1	NM_000612.6

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
IGFALS	100.00	1	NM_004970.3
KDM6A	100.00	1	NM_001291415.2
KMT2D	100.00	1	NM_003482.3
KRAS	100.00	1	NM_033360.4
NPPC	100.00	1	NM_024409.4
NPR2	100.00	1	NM_003995.3
NPR3	100.00	1	NM_001204375.2
NRAS	100.00	1	NM_002524.5
OBSL1	100.00	1	NM_015311.3
PIK3R1	100.00	1	NM_181523.3
POU1F1	100.00	1	NM_000306.4
PTPN11	100.00	1	NM_002834.5
RAF1	100.00	1	NM_001354689.3
RASA2	100.00	1	NM_006506.5
RIT1	100.00	1	NM_006912.6
ROR2	100.00	1	NM_004560.4
RPS6KA3	100.00	1	NM_004586.3
SHOC2	100.00	1	NM_007373.4
SHOX	100.00	1	NM_000451.3
SOS1	100.00	1	NM_005633.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
SOX3	100.00	1	NM_005634.2
SRCAP	100.00	1	NM_006662.3
STAT5B	100.00	1	NM_012448.4
TRIM37	100.00	1	NM_015294.6
WNT5A	100.00	1	NM_003392.4