Supplemental Table S1. Genes selected for variant analysis.

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
ABCC8	NM_000352.6	36890748	Kabuki syndrome with congenital hyperinsulinism and GHD – AD	Diabetes mellitus, noninsulin-dependent (MIM 125853) – AD; Diabetes mellitus, permanent neonatal (MIM 618857) – AD, AR; Hyperinsulinemic hypoglycemia, familial (MIM 256450) – AD,AR; Hypoglycemia of infancy, leucine-sensitive (MIM 240800) – AD
ACAN	NM_001369268.1	34589056	GHD and advanced bone age – AD	?Spondyloepiphyseal dysplasia, Kimberley type (MIM 608361) – AD; Short stature and advanced bone age (MIM 165800) – AD; Spondyloepimetaphyseal dysplasia, aggrecan type (MIM 612813) – AR
ACP5	NM_001111035.3	28740483	Spondyloenchondrodysplasia, with partial GHD – AR	Spondyloenchondrodysplasia with immune dysregulation (MIM 607944) – AR
ADAMTSL2	NM_014694.4	28917829	Geleophysic dysplasia, with GHD and mild motor retardation – AR	Geleophysic dysplasia (MIM 231050) – AR
ADAT3	NM_138422.4	26842963	Intellectual disability syndrome, with brain abnormalities, GHD, and dysmorphic facies – AR	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies (MIM 615286) – AR
AIRE	NM_000383.4	27646917	Autoimmune polyglandular syndrome, with GHD – AD	Autoimmune polyendocrinopathy syndrome (MIM 240300) – AD, AR
AKT3	NM_005465.7	38459620	Megalencephaly, GHD, and central hypothyroidism – AD	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome (MIM 615937) – AD
ALMS1	NM_001378454.1	23445176	Alström syndrome with GHD – AR	Alström syndrome (MIM 203800) – AR
APOB	NM_000384.2	34220717	Familial hypercholesterolemia and GHD – AD	Hypercholesterolemia, familial (MIM 144010) – AD; Hypobetalipoproteinemia (MIM 615558) – AR
ARG1	NM_000045.2	29443755	Argininemia and partial GHD – AR	Argininemia (MIM 207800) – AR
ARID1B	NM_001374820.1	35964110	Coffin-Siris syndrome, GHD, and melanocytic nevi – AD	Coffin-Siris syndrome (MIM 135900) – AD
ARMC9	NM_025139.6	28625504	Joubert syndrome, with GHD, bilateral optic nerve hypoplasia, bifid uvula, and an abnormal brainstem – AR	Joubert syndrome (MIM 617622) – AR
ARNT2	NM_014862.4	24022475	Hypopituitarism (CPHD), post-natal microcephaly, visual and renal anomalies – AR	?Webb-Dattani syndrome (MIM 615926) – AR
ARX	NM_139058.2	15151512	Transsphenoidal encephalocele and CPHD – XLR	Developmental and epileptic encephalopathy (MIM 308350) – XLR; Hydranencephaly with abnormal genitalia (MIM 300215) – XL; Intellectual developmental disorder (MIM 300419) – XLR; Lissencephaly (MIM 300215) – XL; Partington syndrome (MIM 309510) – XLR; Proud syndrome (MIM 300004) – XL
ASH1L	NM_018489.2	32796691	CPHD – AD	Intellectual developmental disorder (MIM 617796) – AD
BLM	NM_000057.4	33122222	Bloom syndrome with GHD – AR	Bloom syndrome (MIM 210900) – AR
BMP4	NM_001202.6	24289245	CPHD and spondyloepiphyseal dysplasia tarda – AD	Microphthalmia, syndromic (MIM 607932) – AD
BPTF	NM_004459.7	36153657	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, with GHD – AD	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies (MIM 617755) – AD
BRAF	NM_004333.6	17551924	Cardiofaciocutaneous syndrome with GHD – AD	Cardiofaciocutaneous syndrome (MIM 115150) – AD; Leopard syndrome (MIM 613707) – AD; Noonan syndrome (MIM 613706) – AD
BRCA1	NM_007294.4	29712865	Neuroblastoma, fanconi anemia and GHD – AR	{Breast–ovarian cancer, familial} (MIM 604370) – AD; Fanconi anemia, complementation group S (MIM 617883) – AR

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
BTK	NM_000061.3	8013627	Agammaglobulinemia and GHD – XLR	Agammaglobulinemia (MIM 300755) – XLR; Isolated growth hormone deficiency with agammaglobulinemia (MIM 307200) – XLR
CACNA1I	NM_021096.4	33704440	Neurodevelopmental disorder with speech impairment and GHD – AD	Neurodevelopmental disorder with speech impairment (MIM 620114) – AD
CDON	NM_016952.5	26529631	Pituitary Stalk Interruption Syndrome with CPHD – AD	Holoprosencephaly (MIM 614226) – AD
CHD7	NM_017780.4	18089695	CHARGE syndrome – AD	Coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities, and ear abnormalities syndrome (MIM 214800) – AD; Hypogonadotropic hypogonadism (MIM 612370) – AD
CLCNKB	NM_000085.3	17185149	Proteinuria, impaired kidney function, and GHD – AR	Bartter syndrome (MIM 607364) – AR; Bartter syndrome (MIM 613090) –DR
COL1A1	NM_000088.4	30984112	Osteogenesis imperfecta, pituitary stalk interruption syndrome, and CPHD – AD	{Bone mineral density variation QTL, osteoporosis} (MIM 166710) – AD; Caffey disease (MIM 114000) – AD; Combined osteogenesis imperfecta and Ehlers–Danlos syndrome (MIM 619115) – AD; Ehlers–Danlos syndrome, arthrochalasia type (MIM 130060) – AD; Osteogenesis imperfecta (MIM 166200, 166210, 259420, 166220) – AD
COL1A2	NM_000089.4	32071780	Osteogenesis imperfecta and CPHD – AD	{Osteoporosis, postmenopausal} (MIM 166710) – AD; Combined osteogenesis imperfecta and Ehlers–Danlos syndrome (MIM 619120) – AD; Ehlers-Danlos syndrome, arthrochalasia type (MIM 617821) – AD; Ehlers-Danlos syndrome, cardiac valvular type (MIM 225320) – AR; Osteogenesis imperfecta (MIM 166210, 259420, 166220) – AD
COL2A1	NM_001844.5	30733658	Spondyloepiphyseal dysplasia congenita and GHD – AD	?Epiphyseal dysplasia, multiple, with myopia and deafness (MIM 132450) – AD; ?Vitreoretinopathy with phalangeal epiphyseal dysplasia (MIM 619248) – AD; Achondrogenesis or hypochondrogenesis (MIM 200610) – AD; Avascular necrosis of the femoral head (MIM 608805) – AD; Czech dysplasia (MIM 609162) – AD; Kniest dysplasia (MIM 156550) – AD; Legg-Calve-Perthes disease (MIM 150600) – AD; Osteoarthritis with mild chondrodysplasia (MIM 604864) – AD; Platyspondylic skeletal dysplasia, Torrance type (MIM 151210) – AD; Spondyloepiphyseal dysplasia congenita (MIM 183900) – AD; Spondyloepimetaphyseal dysplasia, Strudwick type (mim 184250) – AD; Spondyloepiphyseal dysplasia, Stanescu type (MIM 616583) – AD; Spondyloperipheral dysplasia (MIM271700) – AD; Stickler syndrome (MIM 108300) – AD; Stickler syndrome, nonsyndromic ocular (MIM 609508) – AD
COL9A1	NM_001851.4	34826401	GHD and congenital myopia – AD	?Epiphyseal dysplasia, multiple (MIM 614135) – AD; Stickler syndrome (MIM 614134) – AR
COL11A1	NM_001854.4	34589056	Marshall syndrome, depressed nasal bridge, long philtrum, and GHD – AD	Deafness (MIM 618533) – AD; Fibrochondrogenesis (MIM 228520) – AR; Marshall syndrome (MIM 154780) – AD; Stickler syndrome (MIM 604841) – AD
CREBBP	NM_004380.3	23432975	Rubinstein-Taybi syndrome, GHD, and Arnold Chiari malformation – AD	Menke-Hennekam syndrome (MIM 618332) – AD; Rubinstein-Taybi syndrome (MIM 180849) – AD
CSNK2A1	NM_001895.4	38096238	Okur-Chung syndrome, pituitary stalk interruption syndrome, and GHD – AD	Okur-Chung neurodevelopmental syndrome (MIM 617062) – AD
DCC	NM_005215.4	38717911	CPHD with developmental delay – AD	Gaze palsy, familial horizontal, with progressive scoliosis (MIM 617542) – AR; Mirror movements and/or agenesis of the corpus callosum (MIM 157600) – AD

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
DMXL2	NM_001174116.3	38096238	Pituitary stalk interruption syndrome, and GHD – AR	?Deafness, autosomal dominant (MIM 617605) – AD; ?Polyendocrine-polyneuropathy syndrome (MIM 616113) – AR; Developmental and epileptic encephalopathy (MIM 618663) – AR
DNAJC3	NM_006260.5	32738013	Diabetes mellitus, GHD, hypothyroidism and diffuse neurodegeneration – AR	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus (MIM 616192) – AR
DNMT1	NM_001130823.3	33270637	Cerebellar ataxia with pituitary stalk interruption syndrome, and CPHD – AD	Cerebellar ataxia, deafness, and narcolepsy (MIM 604121) – AD; Neuropathy, hereditary sensory (MIM 614116) – AD
DPH1	NM_001383.6	33704902	Loucks-Innes syndrome with GHD – AR	Developmental delay with short stature, dysmorphic facial features, and sparse hair (MIM 616901) – AR
EIF2S3	NM_001415.4	27333055	Syndromic intellectual disability with severe microcephaly, growth retardation (GHD), and epilepsy – XLR	Mental retardation, epileptic seizures, hypogonadism with hypogenitalism, microcephaly, and obesity syndrome (MIM 300148) – XLR
FAM111A	NM_022074.4		Kenny-Caffey syndrome with GHD – AD	Gracile bone dysplasia (MIM 602361) – AD; Kenny-Caffey syndrome (MIM 127000) – AD
FANCA	NM_000135.4	33270637	Fanconi and pituitary stalk interruption syndrome, microftalmia, and CPHD – AR	Fanconi anemia, complementation group A (MIM 227650) – AR
FGF8	NM_033163.5	22319038	CPHD – AD	Hypogonadotropic hypogonadism (MIM 612702) – AD
FGFR1	NM_023110.3	23657145	CPHD and multiple neurological abnormalities – AD	Hartsfield syndrome (MIM 615465) – AD; Hypogonadotropic hypogonadism (MIM 147950) – AD; Jackson-Weiss syndrome (MIM 123150) – AD; Osteoglophonic dysplasia (MIM 166250) – AD; Pfeiffer syndrome (MIM 101600) – AD; Trigonocephaly (MIM 190440) – AD
FGFR3	NM_000142.4	26136890	Achondroplasia and mild GHD – AD	Achondroplasia (MIM 100800) – AD; Camptodactyly, tall stature, and hearing loss syndrome (MIM 610474) – AD, AR; Crouzon syndrome with acanthosis nigricans (MIM 612247) – AD; Hypochondroplasia (MIM 146000) – AD; Lacrimoauriculodentodigital syndrome (MIM 620192) – AD; Muenke syndrome (MIM 602849) – AD; Achondroplasia, severe, with developmental delay and acanthosis nigricans (MIM 616482) – AD; Thanatophoric dysplasia (MIM 187600 and 187601) – AD
FLNB	NM_001457.4	18386804	Spondylocarpotarsal synostosis syndrome with GHD – AR	Atelosteogenesis (MIM 108720 and 108721) – AD; Boomerang dysplasia (MIM 112310) – AD; Larsen syndrome (MIM 150250) – AD; Spondylocarpotarsal synostosis syndrome (MIM 272460) – AR
FOXA2	NM_021784.4	25765999	Heterotaxy, panhypopituitarism (CPHD), and biliary atresia – AD	Unassigned phenotype
FOXL2	NM_023067.3	32454486	Blepharophimosis, ptosis, and epicanthus inversus syndrome with GHD or CPHD – AD	Blepharophimosis, epicanthus inversus, and ptosis (MIM 110100) – AD, AR; Premature ovarian failure (MIM 608996) – AD
FRA10AC1	NM_145246.5	34694367	Neurodevelopmental disorder with clinodactyly, growth retardation and GHD – AR	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities (MIM 620113) – AR
G6PC3	NM_138387.4	22050868	Dursun syndrome with GHD – AR	Neutropenia, severe congenital (MIM 612541) – AR
GH1	NM_000515.5	7962317 (AD) 6273867 (AR)	Isolated GHD – AD, AR	Growth hormone deficiency, isolated, type IA (MIM 262400) – AR; Growth hormone deficiency, isolated, type II (MIM 173100) – AD; Kowarski syndrome (MIM 262650) – AR
GHRHR	NM_000823.4	8528260	Isolated GHD – AR	Growth hormone deficiency, isolated, type IV (MIM 618157) – AR
GHSR	NM_198407.2	16511605	Isolated GHD – AD	Growth hormone deficiency, isolated partial (MIM 615925) – AD, AR

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
GJA1	NM_000165.5	30610049(AD) 14974090(AR)	Oculodentodigital dysplasia with GHD – AD, AR	Craniometaphyseal dysplasia (MIM 218400) – AR; Erythrokeratodermia variabilis et progressiva (MIM 617525) – AD; Oculodentodigital dysplasia (MIM 164200 and 257850) – AD, AR; Palmoplantar keratoderma with congenital alopecia (MIM 104100) – AD; Syndactyly (MIM 186100) – AD
GLI2	NM_005270.5	14581620	Pituitary anomalies (GHD or CPHD) and holoprosencephaly-like features – AD	Culler-Jones syndrome (MIM 615849) – AD; Holoprosencephaly (MIM 610829) – AD
GLI3	NM_000168.6	15739154	Pallister-Hall syndrome with GHD – AD	Greig cephalopolysyndactyly syndrome (MIM 175700) – AD; Pallister-Hall syndrome (MIM 146510) – AD; Polydactyly, postaxial (MIM 174200) – AD; Polydactyly, preaxial (MIM 174700) – AD
GMNN	NM_015895.5	26637980	Meier-Gorlin syndrome with GHD – AD	Meier-Gorlin syndrome (MIM 616835) – AD
GNAO1	NM_020988.3	39078873	CPHD – AD	Developmental and epileptic encephalopathy (MIM 615473) – AD; Neurodevelopmental disorder with involuntary movements (MIM 617493) – AD
GNAS	NM_000516.7	20480732	Progressive osseous heteroplasia and GHD – AD	Osseous heteroplasia, progressive (MIM 166350) – AD; Pseudohypoparathyroidism (MIM 103580, 603233, and 612462) – AD; Pseudopseudohypoparathyroidism (MIM 612463) – AD
GPR161	NM_001267609.1	25322266	Pituitary stalk interruption syndrome and GHD – AR	{Medulloblastoma predisposition syndrome} (MIM 155255) – AD, AR
GRM7	NM_000844.4	32286009	Neurodevelopmental disorder with epilepsy, microcephaly, cerebral atrophy, and GHD or CPHD – AR	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities (MIM 618922) – AR
GYS2	NM_021957.4	11592574	Hepatic glycogen synthase deficiency and GHD – AR	Glycogen storage disease (MIM 240600) – AR
H1-4	NM_005321.3	37362168	Rahman syndrome with CPHD – AD	Rahman syndrome (MIM 617537) – AD
HESX1	NM_003865.3	11136712 (AD) 9620767 (AR)	CPHD with pituitary anomalies and/or septo-optic dysplasia – AD, AR	Growth hormone deficiency with pituitary anomalies (MIM 182230) – AD, AR; Pituitary hormone deficiency, combined (MIM 182230) – AD, AR; Septooptic dysplasia (MIM 182230) – AD, AR
HHIP	NM_022475.3	22897141	CPHD – AD	Unassigned phenotype
HID1	NM_030630.2	28600779	Developmental disorder with CPHD – AR	Developmental and epileptic encephalopathy with hypopituitarism (MIM 619983) – AR
HNF1A	NM_000545.5	21051477	Maturity-onset diabetes of the young, renal dysplasia and CPHD – AD	{Diabetes mellitus, insulin–dependent} (MIM 222100) – AR; {Diabetes mellitus, noninsulin-dependent, 2} (MIM 125853) – AD; Maturity-onset diabetes of the young (MIM 600496) – AD
HNRNPU	NM_031844.3	25590979	Intellectual disability, epilepsy, CPHD, hypertension, and osseus anomalies – AD	Developmental and epileptic encephalopathy (MIM 617391) – AD
HRAS	NM_005343.4	21438134	Costello syndrome with GHD – AD	Congenital myopathy with excess of muscle spindles (MIM 218040) – AD
IARS2	NM_018060.4	25130867	Cataracts, GHD, partial sensorineural deafness, and peripheral neuropathy – AR	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (MIM 616007) – AR
IDS	NM_000202.8	34589056	Mucopolysaccharidosis type II, strabismus, and GHD – XLR	Mucopolysaccharidosis (MIM 309900) – XLR
IFT172	NM_015662.3	25664603	Functional GHD, retinopathy, metaphyseal dysplasia, and hypertension – AR	Bardet-Biedl syndrome (MIM 619471) – AR; Retinitis pigmentosa (MIM 616394) – AR; Short-rib thoracic dysplasia (MIM 615630) – AR
IFT56	NM_001321741.2	38135897	Hexadactyly, pituitary stalk interruption syndrome with CPHD, hepatopathy, nephropathy, and bilateral lip-palate cleft – AR	Biliary, renal, neurologic, and skeletal syndrome (MIM 619534) – AR
IGF2	NM_000612.6	38854326	Russell-Silver syndrome with GHD – AD	Silver-Russell syndrome (MIM 616489) – AD

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
IGSF1	NM_001170961.2	26302767	CPHD – XLR	Hypothyroidism, central, and testicular enlargement (MIM 300888) – XLR
IHH	NM_002181.3	38840672	Short stature and non-classical brachydactyly type A1, and GHD – AD	Acrocapitofemoral dysplasia (MIM 607778) – AR; Brachydactyly (MIM 112500) – AD
KAT6A	NM_006766.5	29165578	Pituitary stalk interruption syndrome with CPHD – AD	Arboleda-Tham syndrome (MIM 616268) – AD
KATNIP	NM_015202.5	32164589	Joubert syndrome with ocular coloboma, pituitary – AR malformation and GHD	Joubert syndrome (MIM 616784) – AR
KCNA2	NM_004974.4	25751627	Epileptic encephalopathy, with ataxia and psychomotor and language development delay, and GHD – AD	Developmental and epileptic encephalopathy (MIM 616366) – AD
KCNQ1	NM_000218.2	29097701	GHD and gingival fibromatosis – AD	{Long QT syndrome, acquired, susceptibility to} (MIM 192500) – AD; Atrial fibrillation, familial (MIM 607554) – AD; Jervell and Lange-Nielsen syndrome (MIM 220400) – AR; Long QT syndrome (MIM 192500) – AD; Short QT syndrome (MIM 609621) – AD
KIAA0753	NM_014804.3	28220259	Joubert syndrome and GHD – AR	?Joubert syndrome (MIM 619476) – AR; ?Orofaciodigital syndrome (MIM 617127) – AR; Short-rib thoracic dysplasia without polydactyly (MIM 619479) – AR
KIF1A	NM_004321.8	25253658	Severe intellectual disability, spasticity, optic atrophy, neurogenic bladder, progressive cerebellar atrophy, and GHD – AD	Neurodegeneration and spasticity with or without cerebellar atrophy or cortical visual impairment syndrome (MIM 614255) – AD; Neuropathy, hereditary sensory (MIM 614213) – AR; Spastic paraplegia (MIM 610357 and 620607) – AD, AR
KISS1R	NM_032551.5	31073722	CPHD – AR	?Precocious puberty, central (MIM 176400) – AD; Hypogonadotropic hypogonadism (MIM 614837) – AR
KMT2A	NM_001197104.2	30159147	Wiedemann-Steiner syndrome with GHD – AD	Wiedemann-Steiner syndrome (MIM 605130) – AD
KMT2D	NM_003482.3	24311525	Kabuki syndrome with GHD – AD	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome (MIM 620186) – AD; Kabuki syndrome (MIM 147920) – AD
L1CAM	NM_000425.5	31504653	Arthrogryposis, and GHD – XLR	?Corpus callosum, partial agenesis of (MIM 304100) – XLR; Hydrocephalus, congenital (MIM 307000) – XLR; Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MIM 303350) – XLR
LAMB2	NM_002292.3	31769495	Albuminuria, optic nerve hypoplasia, and GHD – AR	Nephrotic syndrome (MIM 614199) – AR; Pierson syndrome (MIM 609049) – AR
LDLR	NM_000527.4	34220717	Familial hypercholesterolemia and GHD – AD	Hypercholesterolemia, familial (MIM 143890) – AD, AR
LEPR	NM_002303.6	9537324	Obesity and pituitary dysfunction (CPHD) – AR	Obesity, morbid, due to leptin receptor deficiency (MIM 614963) – AR
LHX3	NM_014564.5	10835633	CPHD – AR	Pituitary hormone deficiency, combined (MIM 221750) – AR
LHX4	NM_033343.4	11567216	CPHD – AD	Pituitary hormone deficiency, combined (MIM 262700) – AD
LIG4	NM_002312.3	39953892	DNA Ligase IV deficiency and GHD – AR	LIG4 syndrome (MIM 606593) – AR
LZTR1	NM_006767.4	33407364 (AD) 29469822(AR)	Noonan syndrome and GHD – AD, AR	{Schwannomatosis, susceptibility to} (MIM 615670) – AD; Noonan syndrome (MIM 616564 and 605275) – AD, AR
MADD	NM_003682.4	32761064	Developmental delay with endocrine, exocrine, autonomic, and hematologic adnormalities syndrome, with GHD – AR	Developmental delay with endocrine, exocrine, autonomic, and hematologic adnormalities syndrome (MIM 619004) – AR; Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia (MIM 619005) – AR
MAGEL2	NM_019066.5	30323850	Schaaf-Yang syndrome with CPHD – AD	Schaaf-Yang syndrome (MIM 615547) – AD
MAP2K1	NM_002755.4	36313893	Cardiofaciocutaneous syndrome with GHD – AD	Cardiofaciocutaneous syndrome (MIM 615279) – AD
MARS2	NM_138395.4	25754315	Developmental delay, sensorineural hearing loss, and GHD – AR	?Combined oxidative phosphorylation deficiency (MIM 616430) – AR; Spastic ataxia (MIM 611390) – AR
MBTPS1	NM_003791.4	36714646	Spondyloepiphyseal dysplasia and GHD – AR	?Spondyloepiphyseal dysplasia, Kondo-Fu type (MIM 618392) – AR

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
MC2R	NM_000529.2	21778684	Familial glucocorticoid deficiency with GHD – AR	Glucocorticoid deficiency, due to ACTH unresponsiveness (MIM 202200) – AR
MMP13	NM_002427.4	34589056	Spondyloepimetaphyseal dysplasia Missouri type, and GHD – AD	Metaphyseal anadysplasia (MIM 602111) – AD; Metaphyseal dysplasia, Spahr type (MIM 250400) – AR
MTFMT	NM_139242.4	24461907	Encephalopathy, lactic acidosis, mitochondrial oxidative phosphorylation defects, and GHD – AR	Combined oxidative phosphorylation deficiency (MIM 614947) – AR; Mitochondrial complex I deficiency, nuclear type (MIM 618248) – AR
MYH11	NM_002474.3	31044419	Megacystis-microcolon-intestinal-hypoperistalsis syndrome, central hypothyroidism, tonically dilated pupil, and GHD – AR	Aortic aneurysm, familial thoracic (MIM 132900) – AD; Megacystis-microcolon-intestinal hypoperistalsis syndrome (MIM 619351) – AR; Visceral myopathy (MIM 619350) – AD
NBAS	NM_015909.2	30592236	Short stature, optic nerve atrophy, and Pelger-Huet anomaly syndrome, and GHD – AR	Infantile liver failure syndrome (MIM 616483) – AR; Short stature, optic nerve atrophy, and Pelger-Huet anomaly (MIM 614800) – AR
NF1	NM_000267.3	26758488	Neurofibromatosis-Noonan syndrome with GHD – AD	Leukemia, juvenile myelomonocytic (MIM 607785) – AD; Neurofibromatosis-Noonan syndrome (MIM 601321) – AD; Neurofibromatosis, familial spinal (MIM 162210) – AD; Neurofibromatosis (MIM 162200) – AD; Watson syndrome (MIM 193520) – AD
NFKBIA	NM_020529.3	23708964	Ectodermal dysplasia, immunodeficiency, polyendocrinopathy with GHD – AD	Ectodermal dysplasia and immunodeficiency (MIM 612132) – AD
NFKB2	NM_001077494.3	25524009	DAVID syndrome, with CPHD – AD	Immunodeficiency, common variable (MIM 615577) – AD
NKX2-1	NM_001079668.2	30186310	Myoclonus, dystonia, and pituitary involvement (CPHD) – AD	{Thyroid cancer, nonmedullary} (MIM 188550) – AD; Chorea, hereditary benign (MIM 118700) – AD; Choreoathetosis, hypothyroidism, and neonatal respiratory distress (MIM 610978) – AD
NOTCH3	NM_000435.3	28707430	Pituitary stalk interruption syndrome with GHD – AD	?Myofibromatosis, infantile (MIM 615293) – AD; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (MIM 125310) – AD; Lateral meningocele syndrome (MIM 130720) – AD
NPHP4	NM_015102.5	34850017	CPHD – AR	Nephronophthisis (MIM 606966) – AR
NPR2	NM_003995.3	36714562	Isolated GHD – AD	Acromesomelic dysplasia, Maroteaux type (MIM 602875) – AR; Epiphyseal chondrodysplasia, Miura type (MIM 615923) – AD; Short stature with nonspecific skeletal abnormalities (MIM 616255) – AD
NR0B1	NM_000475.4	27110597	Adrenal hypoplasia congenita and GHD – XLR	46XY sex reversal, dosage-sensitive (MIM 300018) – XL; Adrenal hypoplasia, congenital (MIM 300200) – XLR
NRAS	NM_002524.5	19966803	Noonan syndrome with partial GHD – AD	Noonan syndrome (MIM 613224) – AD
NSMF	NM_015537.5	33270637	Pituitary stalk interruption syndrome with CPHD – AD	Hypogonadotropic hypogonadism (MIM 614838) – AD
OBSL1	NM_015311.2	33728303	Isolated GHD – AR	3M syndrome (MIM 612921) – AR
OTX2	NM_172337.3	18628516	Anophthalmia, short stature, and partial GHD – AD	Microphtalmia, syndromic (MIM 610125) – AD; Pituitary hormone deficiency, combined (MIM 613986) – AD
PAX6	NM_000280.5	25342853	Cleft palate, optic disc cupping, and GHD – AD	?Coloboma of optic nerve (MIM 120430) – AD; Anterior segment dysgenesis, multiple subtypes (MIM 604229) – AD; Cataract with late-onset corneal dystrophy (MIM 106210) – AD; Forveal hypoplasia (MIM 136520) – AD; Keratitis (MIM 148190) – AD; Microphthalmia/coloboma (MIM 120200) – AD; Optic nerve hypoplasia (MIM 165550) – AD
PAX7	NM_002584.3	28779497	Failure to thrive, hypotonia, and global neurodevelopmental delay syndrome, with GHD – AR	Congenital myopathy (MIM 618578) – AR

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
PCSK1	NM_000439.5	23562752	Malabsorptive diarrhea and CPHD – AR	Endocrinopathy due to proprotein convertase 1/3 deficiency (MIM 600955) – AR
PGM1	NM_002633.3	22976764	Congenital disorder of glycosylation and GHD – AR	Congenital disorder of glycosylation (MIM 614921) – AR
PHF6	NM_032458.3	14714754	Borjeson-Forssman-Lehmann syndrome and CPHD – XLR	Borjeson-Forssman-Lehmann syndrome (MIM 301900) – XLR
PIK3CA	NM_006218.4	31729162	Megalencephaly-capillary malformation syndrome and GHD – AD	Cowden syndrome (MIM 615108)
PIK3CD	NM_005026.5	35189965	Immunodeficiency, bronchiectasis, nephromegaly, and GHD – AD	?Roifman-Chitayat syndrome (MIM 613328) – DR; Immunodeficiency (MIM 615513 and 619281) – AD, AR
PITX1	NM_002653.4	38096238	Pituitary stalk interruption syndrome, with GHD – AD	Clubfoot, congenital (MIM 119800) – AD
PITX2	NM_153427.2	38096238	Pituitary stalk interruption syndrome, with GHD – AD	Anterior segment dysgenesis (MIM 137600) – AD; Axenfeld-Rieger syndrome (MIM 180500) – AD; Ring dermoid of cornea (MIM 180550) – AD
PMM2	NM_000303.3	39078873	CPHD – AR	Congenital disorder of glycosylation (MIM 212065) – AR
PNPLA6	NM_006702.5	25574898	Oliver McFarlane syndrome with GHD – AR	?Laurence-Moon syndrome (MIM 245800) – AR; Boucher-Neuhauser syndrome (MIM 215470) – AR; Oliver-McFarlane syndrome (MIM 275400) – AR; Spastic paraplegia (MIM 612020) – AR
POLR3A	NM_007055.4	22451160	Hypomyelination with hypogonadotropic hypogonadism and hypodontia syndrome with CPHD – AR	Leukodystrophy, hypomyelinating (MIM 607694) – AR; Wiedemann-Rautenstrauch syndrome (MIM 264090) – AR
POLR3B	NM_018082.6	26204956	Hypomyelination with hypogonadotropic hypogonadism and hypodontia syndrome with CPHD – AR	Charcot-Marie-Tooth disease, demyelinating (MIM 619742) – AD; Leukodystrophy, hypomyelinating (MIM 614381) – AR
POMC	NM_001035256.3	18765507	Obesity, adrenal insufficiency, and CPHD – AR	{Obesity, early–onset, susceptibility to} (MIM 601665) – AD, AR; Obesity, adrenal insufficiency, and red hair due to POMC deficiency (MIM 609734) – AR
POU1F1	NM_000306.4	1509262 (AD) 1302000 (AR)	CPHD – AD, AR	Pituitary hormone deficiency, combined or isolated (MIM 613038) – AD, AR
PREPL	NM_006036.4	32218803	Congenital myasthenic syndrome, ovaries absence, utures hypoplasia, and CPHD – AR	Myasthenic syndrome, congenital (MIM 616224) – AR
PRMT7	NM_019023.5	36348013	Short stature (GHD), brachydactyly, intellectual developmental disability, and seizures – AR	Short stature, brachydactyly, intellectual developmental disability, and seizures (MIM 617157) – AR
PROK2	NM_001126128.2	29165578	Pituitary stalk interruption syndrome, with CPHD – AD	Hypogonadotropic hypogonadism (MIM 610628) – AD
PROKR2	NM_144773.4	22319038	CPHD and septo-optic dysplasia – AD	Hypogonadotropic hypogonadism (MIM 244200) – AD
PROP1	NM_006261.5	9462743	CPHD – AR	Pituitary hormone deficiency, combined (MIM 262600) – AR
PRPF8	NM_006445.4	38976971	Hypophosphatemia with delayed puberty and GHD – AD	Retinitis pigmentosa (MIM 600059) – AD
PSEN1	NM_000021.3	28707430	Pituitary stalk interruption syndrome, with GHD – AD	?Acne inversa, familial (MIM 613737) – AD; ?Cardiomyopathy, dilated (MIM 613694) – AD; Alzheimer disease (MIM 607822)– AD; Dementia, frontotemporal (MIM 600274) – AD; Pick disease (MIM 172700) – AD
PTPN11	NM_002834.5	26444854	Noonan syndrome and GHD – AD	Multiple lentigines syndrome (MIM 151100) – AD; Metachondromatosis (MIM 156250) – AD; Noonan syndrome (MIM 163950) – AD
PUS1	NM_025215.6	17056637	Myopathy, lactic acidosis, sideroblastic anemia, and GHD – AR	Myopathy, lactic acidosis, and sideroblastic anemia (MIM 600462) – AR
RAB3GAP1	NM_012233.2	25332050	Warburg micro syndrome with GHD – AR	Martsolf syndrome (MIM 619420) – AR; Warburg micro syndrome (MIM 600118) – AR
RAC2	NM_002872.5	25512081	Common variable immunodeficiency, glomerulonephritis, coagulopathy, and CPHD – AD	?Immunodeficiency with defective neutrophil chemotaxis and hypogammaglobulinemia (MIM 618987) – AR; Immunodeficiency with defective

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
				neutrophil chemotaxix and leukocytosis (MIM 608203) – AD; Immunodeficiency with defective neutrophil chemotaxis and lymphopenia (MIM 618986) – AD
RAF1	NM_002880.4	33270637	Pituitary stalk interruption syndrome, deafness, and GHD – AD	Cardiomyopathy, dilated (MIM 615916) – AD; Multiple lentigines syndrome (MIM 611554) – AD; Noonan syndrome (MIM 611553) – AD
RAX	NM_013435.2	30811539	Anophthalmia, hypopituitarism (CPHD), diabetes insipidus, and cleft palate – AR	Microphthalmia, syndromic (MIM 611038) – AR
RBM28	NM_018077.3	20231366	Alopecia, neurological defects, and endocrinopathy syndrome, with CPHD – AR	?Alopecia, neurologic defects, and endocrinopathy syndrome (MIM 612079) – AR
RECQL4	NM_004260.4	17372760	Rothmund-Thomson syndrome and GHD – AR	Baller-Gerold syndrome (MIM 218600) – AR; Short stature, radial ray defects and other malformations, and infantile diarrhea syndrome (MIM 266280) – AR; Rothmund-Thomson syndrome (MIM 268400) – AR
RFWD3	NM_018124.4	28691929	Fanconi anemia and partial GHD – AR	?Fanconi anemia, complementation group W (MIM 617784) – AR
RNPC3	NM_017619.4	24480542	Isolated GHD – AR	Pituitary hormone deficiency, combined or isolated (MIM 618160) – AR
ROBO1	NM_002941.4	28402530	Pituitary stalk interruption syndrome, ocular anomalies, and GHD/CPHD – AD	?Nystagmus, congenital (MIM 257400) – AR; Neurooculorenal syndrome (MIM 620305) – AR; Pituitary hormone deficiency, combined or isolated (MIM 620303) – AD
ROR2	NM_004560.4	34589056	Brachydactyly type B1, and GHD – AD	Brachydactyly (MIM 113000) – AD; Robinow syndrome (MIM 268310) – AR
SALL4	NM_020436.3	36714562	GHD, radial ray defect, and kidney dystopia – AD	?IVIC syndrome (MIM 147750) – AD; Duane-radial ray syndrome (MIM 607323) – AD
SCO1	NM_004589.4	39214134	Mitochondrial complex IV deficiency nuclear type 4, with developmental and epileptic encephalopathy, and CPHD – AR	Mitochondrial complex IV deficiency, nuclear type (MIM 619048) – AR
SEMA3A	NM_006080.3	31667184	Pituitary (CPHD), heart, kidney, and skeletal dysplasia – AD	{Hypogonadotropic hypogonadism} (MIM 614897) – AD
SHH	NM_000193.4	22897141	CPHD – AD	Holoprosencephaly (MIM 142945) – AD; Microphthalmia with coloboma (IM 611638) – AD; Single median maxillary central incisor (MIM 147250) – AD
SHOC2	NM_007373.4	22995099	Noonan-like syndrome with loose anagen hair and GHD – AD	Noonan syndrome-like with loose anagen hair (MIM 607721) – AD
SHOX	NM_000451.3	38956755	Leri-Weill dyschondrosteosis and GHD – PD	Langer mesomelic dysplasia (MIM 249700) – PR; Leri-Weill dyschondrosteosis (MIM 127300) – PD
SIX3	NM_005413.4	29333838	Solitary median maxillary central incisor, holoprosencephaly, congenital nasal pyriform aperture stenosis, and CPHD – AD	Holoprosencephaly (MIM 157170) – AD
SLC7A7	NM_001126106.4	16775724	Lysinuric protein intolerance and GHD – AR	Lysinuric protein intolerance (MIM 222700) – AR
SLC12A6	NM_133647.1	29165578	Pituitary stalk interruption syndrome with CPHD – AD	Agenesis of the corpus callosum with peripheral neuropathy (MIM 218000) – AR; Charcot-Marie-Tooth disease, axonal (MIM 620068) – AD
SLC20A1	NM_005415	29261175	CPHD – AR	Unassigned phenotype
SLC26A2	NM_000112.3	33728303	Isolated GHD – AR	Achondrogenesis (MIM 600972) – AR; De la Chapelle dysplasia (MIM 256050) – AR; Diastrophic dysplasia (MIM 222600) – AR; Epiphyseal dysplasia, multiple (MIM 226900) – AR
SLC29A3	NM_018344.6	30517079	Histiocytosis-lymphadenopathy plus syndrome, with short stature (GHD) and diabetes mellitus – AR	Histiocytosis-lymphadenopathy plus syndrome (MIM 602782) – AR
SMARCA2	NM_003070.3	33270637	Pituitary stalk interruption syndrome, epilepsy, severe intellectual deficiency, and CPHD – AD	Blepharophimosis-impaired intellectual development syndrome (MIM 619293) – AD; Nicolaides-Baraitser syndrome (MIM 601358) – AD
SON	NM_032195.3	32926520	Zhu-Tokita-Takenouchi-Kim syndrome and GHD – AD	Zhu-Tokita-Takenouchi-Kim syndrome (MIM 617140) – AD

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
SOX2	NM_003106.4	16932809	Bilateral eye defects, other variable defects, and CPHD – AD	Optic nerve hypoplasia and abnormalities of the central nervous system (MIM 206900) – AD
SOX3	NM_005634.3	12428212	Mental retardation and GHD – XL	Panhypopituitarism (MIM 312000) – XL
STAR	NM_000349.2	33536409	Lipoid adrenal hyperplasia and CPHD – AR	Lipoid adrenal hyperplasia (MIM 201710) – AR
STAT5B	NM_012448.4	34589056	Growth hormone insensitivity with immunodeficiency, and GHD – AD	Growth hormone insensitivity with immune dysregulation (MIM 618985 and 245590) – AD, AR
STX16	NM_001001433.3	25843330	Pseudohypoparathyroidism type Ib and GHD – AD	Pseudohypoparathyroidism Ib (MIM 603233) – AD
TAB2	NM_015093.6	33728303	Isolated GHD – AR	Congenital heart defects, nonsyndromic (MIM 614980) – AD
TBC1D32	NM_152730.6	24285566	Orofaciodigital syndrome and CPHD– AR	Unassigned phenotype
TBCE	NM_003193.5	19491227	Hypoparathyroidism-retardation-dysmorphism syndrome with CPHD – AR	Encephalopathy, progressive, with amyotrophy and optic atrophy (MIM 617207) – AR; Hypoparathyroidism-retardation-dysmorphism syndrome (MIM 241410) – AR; Kenny-Caffey syndrome (MIM 244460) – AR
TBX2	NM_005994.4	29726930	Syndromic cardiovascular and skeletal developmental disorder with GHD – AD	Vertebral anomalies and variable endocrine and T-cell dysfunction (MIM 618223) – AD
TBX3	NM_005996.4	19938096	Ulnar-mammary syndrome with GHD – AD	Ulnar-mammary syndrome (MIM 181450) – AD
TCF7L1	NM_031283.3	26764381	Septo-optic dysplasia and GHD – AD	Unassigned phenotype
TGFBR2	NM_003242.6	39830008	Loeys–Dietz syndrome and GHD – AD	Loeys-Dietz syndrome (MIM 610168) – AD
TGIF1	NM_173208.3	23476075	Pituitary stalk interruption syndrome, single central incisor, and CPHD – AD	Holoprosencephaly (MIM 142946) – AD
TMEM67	NM_153704.6	29891882	RHYNS syndrome with CPHD – AR	?Retinitis pigmentosa syndrome (MIM 602152) – AR; {Bardet–Biedl syndrome, modifier of} (MIM 615991) – AR; Cerebellar vermis hypo/aplasia, oligophrenia, ataxia, ocular coloboma, and hepatic fibrosis syndrome (MIM 216360) – AR; Joubert syndrome (MIM 610688) – AR; Meckel syndrome (MIM 607361) – AR; Nephronophthisis (MIM 613550) – AR
TRMT10A	NM_152292.5	34541035	Intellectual disability, microcephaly, short stature, and diabetes syndrome with GHD – AR	Microcephaly, short stature, and impaired glucose metabolism (MIM 616033) – AR
TRPS1	NM_014112.5	22964620	Trichorhinophalangeal syndrome type I – AD	Trichorhinophalangeal syndrome (MIM 190350 and 190351) – AD
WDR4	NM_033661.5	26416026	Distinct facial dysmorphism, brain malformation, severe encephalopathy, and GHD – AR	Galloway-Mowat syndrome (MIM 618347) – AR; Microcephaly, growth deficiency, seizures, and brain malformations (MIM 618346) – AR
WDR11	NM_018117.12	25064402	CPHD – AD	Hypogonadotropic hypogonadism (MIM 614858) – AD; Intellectual developmental disorder (MIM 620237) – AR
WT1	NM_024426.6	33270637	Pituitary stalk interruption syndrome with GHD – AD	Denys-Drash syndrome (MIM 194080) – AD; Frasier syndrome (MIM 136680) – AD; Meacham syndrome (MIM 608978) – AD; Nephrotic syndrome (MIM 256370) – AD; Wilms tumor (MIM 194070) – AD
ZIC2	NM_007129.5	24706429	Holoprosencephaly and panhypopituitarism (CPHD) – AD	Holoprosencephaly (MIM 609637) – AD
ZNF148	NM_021964.3	27964749	Corpus callosum defects, developmental delay, short stature, and dysmorphisms, with GHD – AD	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies (MIM 617260) – AD
ZNF462	NM_021224.6	33975400	Weiss-Kruszka syndrome, with GHD – AD	Weiss-Kruszka syndrome (MIM 618619) – AD

Gene symbol	Reference transcript	First report of association with GHD (PMID)	Phenotype and mode of inheritance in the first report of association with GHD	Phenotypes and modes of inheritance in OMIM
ZSWIM6	NM_020928.2	25105228	Acromelic frontonasal dysostosis and GHD – AD	Acromelic frontonasal dysostosis (MIM 603671) – AD; Neurodevelopmental disorder with movement adnormalities, abnormal gait, and autistic features (MIM 617865) – AD

GHD, Growth Hormone Deficiency; PMID, PubMed identifier; AD, autosomal dominant; AR, autosomal recessive; CPHD, Combined Pituitary Hormone Deficiency; XLR, X–linked recessive; CHARGE, Coloboma, Heart defects, Atresia choanae, growth Retardation, Genital abnormalities, and Ear abnormalities; PD, pseudoautosomal dominant; XL, X–linked; RHYNS, Retinitis pigmentosa, HYpopituitarism, Nephronophthisis and Skeletal dysplasia; OMIM, Online Mendelian Inheritance in Man; MIM, Mendelian Inheritance in Man identifier; DR, digenic recessive; PR, pseudoautosomal recessive.

Supplemental Table S2. Characteristics of 172 GH deficiency patients studied by exome sequencing of 184 genes (only P, LP and VUS variants are presented).

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
2343	M	GH, LH/FSH, TSH, ACTH	-	Hypo, PSI, EN	NM_021096.4(*CACNA1I*):c.1963G>A (p.Val655Ile)	Het	0.000007	0	VUS (PM2, PP2)	-
2493	F	GH, LH/FSH, PRL, ADH	-	ES	NM_014564.5(*LHX3*):c.160C>A (p.Arg54Ser)	Het	0	0	VUS (PM2, PP3)	-
					NM_004260.4(*RECQL4*):c.1793C>T (p.Pro598Leu)	Het	0.000002	0	VUS (PM2)	-
					NM_020436.3(*SALL4*):c.586C>T (p.Arg196Trp)	Het	0.000058	0	VUS (PM2)	19619907
2553	M	GH, LH/FSH, TSH, ACTH	-	N	none	-	-	-	-	-
2610	M	GH, LH/FSH, TSH, ACTH	-	ES, Hypo, EN	NM_021096.4(*CACNA1I*):c.3043G>C (p.Ala1015Pro)	Het	0.000091	0	VUS (PM2, PP2, BS2)	-
					NM_005270.5(*GLI2*):c.1612G>A (p.Glu538Lys)	Het	0.000002	0	VUS (PM2)	-
					NM_004321.8(*KIF1A*):c.2281C>T (p.Pro761Ser)	Het	0	0	VUS (PM2, PP2)	-
					NM_001035256.3(*POMC*):c.286G>T (p.Gly96Cys)	Het	0.000138	0	VUS (PM2)	18091355
					NM_004260.4(*RECQL4*):c.1151G>A (p.Arg384Gln)	Het	0.000710	0	VUS (PM2, BP6)	-
					NM_002941.4(*ROBO1*):c.2954A>G (p.Asn985Ser)	Het	0.000013	0	VUS (PM2)	-
2612	F	GH	Pubertal delay	Hypo, EN	NM_017780.4(*CHD7*):c.7727A>T (p.Asp2576Val)	Het	0	0	VUS (PM2, PP2)	23533228
					NM_017780.4(*CHD7*):c.8338G>A (p.Ala2780Thr)	Het	0.000001	0	VUS (PM2, PP2, BP4)	-
					NM_000545.5(*HNF1A*):c.514G>C (p.Glu172Gln)	Het	0	0	VUS (PM1, PM2, PP3)	-
					NM_015311.2(*OBSL1*):c.3682C>T (p.Arg1228Ter)	Het	0.000004	0	LP (PVS1, PM2)	-
					NM_007055.4(*POLR3A*):c.3718G>A (p.Gly1240Ser)	Het	0.000008	0	VUS (PM2, PP2, PP3)	25339210
2614	M	GH	-	Hypo, PSI, EN	NM_001111035.3(*ACP5*):c.766G>C (p.Val256Leu)	Het	0.000903	0	VUS	-
					NM_015895.5(*GMNN*):c.136G>A (p.Ala46Thr)	Het	0.000039	0	VUS (PM2, BP4)	-
					NM_021957.4(*GYS2*):c.1774C>G (p.Leu592Val)	Het	0.000207	0.002	VUS (PM2)	-
					NM_003482.3(*KMT2D*):c.2264_2317delGGCCTGAGGAGCCACACCTATCTCCGCAGGCTGAGGAGCCACACCTGTCCCCCC (p.Arg755_Pro772del)	Het	0.000005	0	VUS (PM2, PM4, BP6)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_003482.3(*KMT2D*):c.9490C>T (p.Arg3164Trp)	Het	0.000013	0	VUS (PM2, PP2, BP6)	-
					NM_003791.4(*MBTPS1*):c.1130C>G (p.Thr377Ser)	Het	0.000105	0	VUS (PM2)	-
					NM_152730.6(*TBC1D32*):c.2200C>T (p.Arg734Ter)	Het	0.000120	0	LP (PVS1, PM2, PM3)	Doi:10.5734/JGM.2021.18.1.64
2640	M	GH, LH/FSH, TSH	Cryptorchidism	Hypo	NM_005270.5(*GLI2*):c.1738T>C (p.Ser580Pro)	Het	0	0	VUS (PM2, PP3)	-
					NM_002941.4(*ROBO1*):c.1373G>A (p.Arg458Gln)	Het	0.000048	0	VUS (PM2, BP4)	-
2711	M	GH	Cryptorchidism, pubertal delay	N	NM_000384.2(*APOB*):c.10138A>G (p.Lys3380Glu)	Het	0	0	VUS (PM2)	-
					NM_003791.4(*MBTPS1*):c.337G>T (p.Gly113Trp)	Het	0.000001	0	VUS (PM2)	-
					NM_002653.4(*PITX1*):c.691C>A (p.Pro231Thr)	Het	0.000054	0.002	VUS (PM2)	-
					NM_152730.6(*TBC1D32*):c.943G>C (p.Glu315Gln)	Het	0.000096	0.004	VUS (PM2, BP4)	-
2739	F	GH, LH/FSH, TSH, ACTH	-	PSI, RC	NM_001170961.2(*IGSF1*):c.2696G>A (p.Arg899His)	Het	0.000260	0	VUS (PM2, BP4)	-
					NM_004321.8(*KIF1A*):c.4379C>T (p.Thr1460Met)	Het	0.000011	0	VUS (PM2, PP2, BP4)	-
					NM_021224.6(*ZNF462*):c.134G>A (p.Arg45Gln)	Het	0.000001	0	VUS (PM2, PP2, BP4)	-
2763	F	GH, LH/FSH, TSH, ACTH	-	n/a	none	-	-	-	-	-
2806	M	GH	Pubertal delay	N	NM_138422.4(*ADAT3*):c.456_462delGGGCCAG (p.Arg152fsTer19)	Het	0.000007	0.002	LP (PVS1, PM2)	-
					NM_001378454.1(*ALMS1*):c.12251G>T (p.Arg4084Met)	Het	0.000199	0.005	VUS (PM2, BP4)	-
					NM_000061.3(*BTK*):c.1358C>T (p.Ser453Phe)	Hemi	0.000007	0	VUS (PM1, PM2, PP2, PP3, BS2)	-
2808	M	GH, LH/FSH, TSH, PRL, ACTH	-	N	NM_005270.5(*GLI2*):c.3529A>G (p.Lys1177Glu)	Het	0.000006	0	VUS (PM2, BP4)	-
					NM_004260.4(*RECQL4*):c.2351G>A (p.Arg784Gln)	Het	0.000141	0	VUS (PM2)	-
2809	M	GH, LH/FSH, TSH, PRL, ACTH	-	Hypo	NM_000823.4(*GHRHR*):c.976C>T (p.Arg326Cys)	Het	0.000010	0.002	VUS (PM2)	-
2810	M	GH, LH/FSH, TSH, ACTH	-	n/a	NM_001374820.1(*ARID1B*):c.5143G>A (p.Val1715Ile)	Het	0.000007	0	VUS (PM2, PP3)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_014862.4(*ARNT2*):c.1210C>T (p.Arg404Cys)	Het	0.000055	0	VUS (PM2)	-
					NM_015311.2(*OBSL1*):c.3445G>A (p.Gly1149Arg)	Het	0.000035	0	VUS (PM2)	22495311
2813	M	GH, TSH	-	Hypo	NM_198407.2(*GHSR*):c.611C>A (p.Ala204Glu)	Het	0.000032	0	VUS (PM2, PP5)	14715843
					NM_000112.3(*SLC26A2*):c.835C>T (p.Arg279Trp)	Het	0.001659	0.002	LP (PS3, PM1, PM2, PP3, PP5)	8571951
2814	M	GH, TSH, ACTH	Epilepsy, mental retardation	Hypo	NM_001378454.1(*ALMS1*):c.11446C>T (p.Gln3816Ter)	Het	0.000044	0	P (PVS1, PM2, PM3)	11941369
					NM_000135.4(*FANCA*):c.3556A>G (p.Arg1186Gly)	Het	0.000032	0	VUS (PM2)	34598035
					NM_005270.5(*GLI2*):c.3679C>T (p.Gln1227Ter)	**Het**	**0**	**0**	**LP (PVS1, PM2)**	**-**
					NM_002292.3(*LAMB2*):c.110C>T (p.Pro37Leu)	Het	0.0000003	0	VUS (PM2, BP4)	-
					NM_015102.5(*NPHP4*):c.176G>A (p.Arg59Gln)	Het	0.000105	0	VUS (PM2, PP3)	-
					NM_152730.6(*TBC1D32*):c.877C>T (p.Arg293Cys)	Het	0.000577	0.007	VUS	-
2815	M	GH, LH/FSH, TSH, ACTH	-	Hypo	NM_014694.4(*ADAMTSL2*):c.1477G>A (p.Glu493Lys)	Het	0.000359	0	VUS (PM2)	-
2816	M	GH, LH/FSH, TSH, ACTH	-	Hypo, EN	NM_001378454.1(*ALMS1*):c.1852A>G (p.Thr618Ala)	Het	0.000010	0	VUS (PM2, BP4)	-
2817	M	GH, LH/FSH, TSH	-	N	NM_000135.4(*FANCA*):c.3427C>G (p.Leu1143Val)	Het	0.000496	0.002	VUS (PM2, BP6)	14695169
					NM_015909.2(*NBAS*):c.3899C>T (p.Ala1300Val)	Het	0	0	VUS (PM2)	-
					NM_032195.3(*SON*):c.4880_4882delATG (p.Asp1627del)	Het	0.000045	0	VUS (PM2, PM4, BP6)	-
2818	M	GH, LH/FSH, TSH, ACTH	-	Hypo, PSI	NM_015662.3(*IFT172*):c.2953G>A (p.Gly985Ser)	Het	0.000108	0	VUS (PM2)	-
					NM_032551.5(*KISS1R*):c.412G>C (p.Val138Leu)	Het	0.000004	0	VUS (PM2, BP4)	-
					NM_004560.4(*ROR2*):c.2240G>A (p.Arg747Gln)	Het	0.000224	0	VUS (PM2, BS2)	-
2929	M	GH	Cryptorchidism, kidney malformation	N	NM_001895.4(*CSNK2A1*):c.128G>A (p.Arg43Gln)	Het	0.000001	0.002	VUS (PM1, PM2, PP2)	-
					NM_021224.6(*ZNF462*):c.2474A>G (p.Tyr825Cys)	Het	0.000054	0.002	VUS (PM2, PP2, BS2)	-
3004	F	GH, ACTH	-	Hypo	DEL:chr17:63917209-63918917 (1.7 Kb, including part of *GH1*) [§]	Het	-	-	P (1.00)	-
3042	M	GH, LH/FSH, ACTH	-	Hypo	NM_001267609.1(*GPR161*):c.1166G>A (p.Gly389Asp)	Het	0.000006	0	VUS (PM2)	-
					NM_019066.5(*MAGEL2*):c.1180G>A (p.Val394Ile)	Het	0	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
3044	M	GH, LH/FSH	Neonatal seizures and intracerebral hemorrhage	Hypo, PSI	NM_000352.6(*ABCC8*):c.3158G>A (p.Ser1053Asn)	Het	0.000009	0	VUS (PM2, PP2)	-
					DUP:chr17:63873099-63918412 (45.3 Kb, including part of *GH1*) §	-	-	-	VUS (-0.42)	-
					NM_002834.5(*PTPN11*):c.1678C>T (p.Leu560Phe)	Het	0.000115	0.002	VUS (PP2)	12960218
3195	F	GH, LH/FSH, TSH, ACTH	-	ES	NM_005270.5(*GLI2*):c.2265C>G (p.Asn755Lys)	Het	0.000002	0	VUS (PM2, BP4)	-
					NM_002941.4(*ROBO1*):c.979T>C (p.Ser327Pro)	Het	0.000312	0	VUS (PM2, BS2)	35534675
3282	M	GH	-	ES, Hypo, EN	NM_004459.7(*BPTF*):c.5265_5267delTAC (p.Thr1756del)	Het	0.000354	0.002	VUS (PM2, PM4, BP6)	-
					NM_000527.4(*LDLR*):c.2231G>A (p.Arg744Gln)	Het	0.001009	0	VUS (BP4)	9409298
3318	M	GH, LH/FSH	-	Hypo	NM_001111035.3(*ACP5*):c.791T>A (p.Met264Lys)	Het	0.000006	0	LP (PS3, PM2, PM3)	21217752
					NM_021096.4(*CACNA1I*):c.4721A>G (p.Asn1574Ser)	Het	0.000018	0	VUS (PM2, PP2)	-
					NM_001174116.3(*DMXL2*):c.8203G>A (p.Ala2735Thr)	Het	0.000023	0	VUS (PM2, BP4)	-
					NM_002292.3(*LAMB2*):c.253G>A (p.Glu85Lys)	Het	0.000430	0.002	VUS (PM2, BP6)	-
					NM_000435.3(*NOTCH3*):c.5684G>T (p.Arg1895Leu)	Het	0	0	VUS (PM2, PP2)	-
3319	M	GH, LH/FSH, TSH, ACTH	-	Hypo, PSI, EN	NM_001384.4(*DPH2*):c.761C>G (p.Thr254Arg)	Het	0	0	VUS (PM2)	-
					NM_005270.5(*GLI2*):c.4240G>C (p.Glu1414Gln)	Het	0.000011	0	VUS (PM2)	-
					NM_006767.4(*LZTR1*):c.1723G>A (p.Asp575Asn)	Het	0.000541	0.002	VUS (BP4)	-
3330	M	GH, LH/FSH, TSH	-	N	none	-	-	-	-	-
3377	M	GH, TSH, ACTH	SOD, epilepsy, hydrocephalus, myopathy	Hypo, EN	NM_198407.2(*GHSR*):c.108G>C (p.Gln36His)	Het	0.000013	0	VUS (PM2, BP4)	-
					NM_015102.5(*NPHP4*):c.2021G>A (p.Arg674His)	Het	0.000225	0	VUS (PM2, PP3)	30143558
					NM_018082.6(*POLR3B*):c.205G>A (p.Ala69Thr)	Het	0	0	VUS (PM2, PP2)	-
3393	F	GH, LH/FSH, TSH, PRL, ACTH, ADH	Optic nerve glioma	n/a	DUP:chr21:33531431-33718904 (187.5 Kb, including *SON*) §	-	-	-	VUS (0.30)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
3413	F	GH, TSH, ACTH, ADH	Neonatal seizures, mental retardation	Hypo	NM_004459.7(*BPTF*):c.4747G>A (p.Val1583Ile)	Het	0.000004	0	VUS (PM2, PP2, BP4)	-
					NM_004459.7(*BPTF*):c.4752A>T (p.Lys1584Asn)	Het	0	0	VUS (PM2, PP2)	-
					NM_015311.2(*OBSL1*):c.4825G>C (p.Val1609Leu)	Het	0.000001	0	VUS (PM2, BP4)	-
					NM_006445.4(*PRPF8*):c.3515A>G (p.Asn1172Ser)	Het	0	0	VUS (PM2, PP2)	-
					NM_004589.4(*SCO1*):c.307G>A (p.Ala103Thr)	Homo	0.0000003	0	VUS (PM2, PM3)	-
5042	F	GH	-	Hypo, PSI	NM_002312.3(*LIG4*):c.1739G>A (p.Arg580Gln)	Het	0.001036	0	VUS (PP3)	27104957
5044	M	GH	Cleft palate and lip, micropenis, hydrocephalus, hypertrophic myocardiopathy	N	**NM_003482.3(*KMT2D*):c.6595delT (p.Tyr2199fsTer65)**	**Het**	**0**	**0**	**P (PVS1, PS2, PM2, PP5)**	**20711175**
					NM_020928.2(*ZSWIM6*):c.2557G>A (p.Glu853Lys)	Het	0.000007	0	VUS (PM2, PP2)	-
5046	M	GH, LH/FSH, TSH, ACTH	-	Hypo, PSI, EN	none	-	-	-	-	-
5047	M	GH, LH/FSH, ACTH	-	Hypo, PSI, EN	NM_004459.7(*BPTF*):c.6614G>A (p.Arg2205Gln)	Het	0.000429	0.002	VUS (PM2, PP2)	-
					NM_003242.6(*TGFBR2*):c.227T>C (p.Ile76Thr	Het	0	0	VUS (PM2, PP2)	-
5049	F	GH, LH/FSH	-	n/a	NM_001111035.3(*ACP5*):c.386G>A (p.Arg129His)	Het	0.000054	0.002	VUS (PM2, PP3)	-
					NM_016952.5(*CDON*):c.3276+1G>T	**Het**	**0.000309**	**0**	**P (PVS1, PS3, PM2, BS2)**	**-**
					NM_001844.5(*COL2A1*):c.1523A>G (p.Glu508Gly)	Het	0.000008	0	VUS (PM1, PM2, PP2, PP3)	-
					DEL:chr17:63917209-63918917 (1.7 Kb, including part of *GH1*) [§]	Het	-	-	P (1.00)	-
					NM_032551.5(*KISS1R*):c.872C>T (p.Ala291Val)	Het	0.000032	0.004	VUS (PM2, BP4)	-
					NM_015311.2(*OBSL1*):c.4720C>T (p.Arg1574Trp)	Het	0.000134	0	VUS (PM2)	-
5053	M	GH, LH/FSH, TSH, ACTH	Neonatal hypoglycemia, SOD	n/a	**NM_001844.5(*COL2A1*):c.2471G>A (p.Arg824His)**	**Het**	**0.000003**	**0**	**LP (PM1, PM2, PP2, PP3)**	**-**
					NM_012233.2(*RAB3GAP1*):c.2837C>T (p.Pro946Leu)	Het	0.000053	0	VUS (PM2)	-
					NM_002872.5(*RAC2*):c.364G>A (p.Asp122Asn)	Het	0.0000003	0	VUS (PM2, BP6)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_004260.4(*RECQL4*):c.2008G>A (p.Val670Ile)	Het	0.000008	0.002	VUS (PM2)	-
5057	M	GH, TSH, ACTH	Neonatal hypoglycemia, SOD	Hypo, EN	none	-	-	-	-	-
5058	F	GH	Cleft palate, mental retardation, coarctation of the aorta, unilateral renal agenesis	PSI, EN	**NM_003482.3(*KMT2D*):c.13159C>T (p.Gln4387Ter)**	**Het**	**0**	**0**	**LP (PVS1, PM2)**	**25755104**
					NM_002941.4(*ROBO1*):c.4928A>G (p.Asn1643Ser)	Het	0.000002	0	VUS (PM2, BP4)	-
5059	M	GH, LH/FSH	-	Hypo	NM_001369268.1(*ACAN*):c.1189G>A (p.Val397Met)	Het	0.000101	0	VUS (PM2, BP4)	-
					NM_005270.5(*GLI2*):c.547G>A (p.Val183Met)	Het	0.000014	0	VUS (PM2, BP4)	24744436
					NM_030630.2(*HID1*):c.490C>T (p.Arg164Trp)	Het	0.000416	0	VUS (PM2, BS2)	-
					NM_002427.4(*MMP13*):c.662C>T (p.Ala221Val)	Het	0.001641	0	VUS (BS1)	-
					NM_015311.2(*OBSL1*):c.2990C>T (p.Pro997Leu)	Het	0.000014	0	VUS (PM2, BP4)	-
					NM_018077.3(*RBM28*):c.526A>G (p.Met176Val)	Het	0.000359	0	VUS (PM2)	-
5061	M	GH	-	N	NM_004459.7(*BPTF*):c.7741A>T (p.Ser2581Cys)	Het	0.000011	0	VUS (PM2, PP2)	-
5062	M	GH	-	N	NM_004380.3(*CREBBP*):c.7171A>G (p.Met2391Val)	Het	0.000007	0	VUS (PM2, PP2, PP3)	-
					NM_003482.3(*KMT2D*):c.10185_10202dupATTGGCAATGCAGCAGCA (p.Met3398_Ala3403dup)	Het	0.000044	0	VUS (PM2, PM4)	-
					NM_004260.4(*RECQL4*):c.2758A>G (p.Ile920Val)	Het	0.000008	0.002	VUS (PM2)	-
5120	M	GH, LH/FSH	-	Hypo, EN	none	-	-	-	-	-
5134	F	GH	-	N	NM_001378454.1(*ALMS1*):c.2893T>C (p.Tyr965His)	Het	0.000232	0	VUS (PM2, BP4)	-
					NM_001854.4(*COL11A1*):c.1120G>A (p.Val374Ile)	Het	0	0	VUS (PM2)	-
					NM_001854.4(*COL11A1*):c.1121T>A (p.Val374Glu)	Het	0	0	VUS (PM2)	-
					NM_018077.3(*RBM28*):c.708_710delTGA (p.Asp237del)	Het	0.000102	0	VUS (PM2, BP3)	-
5137	M	GH, TSH	-	N	NM_004321.8(*KIF1A*):c.4865C>T (p.Ala1622Val)	Het	0.000493	0.005	VUS (PP2, BP6)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_001126128.2(*PROK2*):c.297dupT (p.Gly100TrpfsTer22)	**Het**	**0.000265**	**0**	**LP (PVS1, PM2)**	**17054399**
5138	F	GH	-	N	NM_018077.3(*RBM28*):c.160A>G (p.Met54Val)	Het	0.000015	0	VUS (PM2)	-
5141	M	GH	-	N	NM_000515.5(*GH1*):c.167A>T (p.Glu56Val)	Het	0.000006	0	VUS (PM2, PP3)	-
5142	F	GH	-	N	NM_015102.5(*NPHP4*):c.203G>A (p.Arg68Gln)	Het	0.000189	0	VUS (PM2)	-
5143	F	GH, TSH, ACTH, ADH	Single central incisor	Hypo, PSI, EN	NM_018489.2(*ASH1L*):c.1070A>G (p.Lys357Arg)	Het	0.000003	0	VUS (PM2, PP2)	-
					DUP:chr16:89816428-89932770 (116.3 Kb, including part of *FANCA*) [§]	-	-	-	VUS (0.30)	-
					NM_005270.5(*GLI2*):c.1484G>A (p.Arg495Gln)	Het	0	0	VUS (PM2)	-
					NM_014804.3(*KIAA0753*):c.1946C>G (p.Ser649Cys)	Het	0	0	VUS (PM2)	-
					DUP:chr2:240682208-240769881 (87.7 Kb, including part of *KIF1A*) [§]	-	-	-	VUS (-0.25)	-
					NM_018077.3(*RBM28*):c.708_710delTGA (p.Asp237del)	Het	0.000102	0	VUS (PM2, BP3)	-
					NM_032195.3(*SON*):c.3581C>A (p.Thr1194Asn)	Het	0	0	VUS (PM2, BP4)	-
					NM_152292.5(*TRMT10A*):c.899G>A (p.Gly300Asp)	Het	0.000180	0	VUS (PM2, BP4)	-
5144	M	GH, TSH, ACTH, ADH	SOD, mental retardation	n/a	NM_001267609.1(*GPR161*):c.1229C>T (p.Thr410Met)	Het	0.000015	0	VUS (PM2)	-
5145	M	GH	-	N	NM_015662.3(*IFT172*):c.5053C>T (p.Pro1685Ser)	Het	0.000054	0	VUS (PM2)	-
5162	M	GH, LH/FSH	-	N	NM_000425.5(*L1CAM*):c.751C>G (p.His251Asp)	Hemi	0	0	VUS (PM2)	-
5165	M	GH	-	N	NM_001378454.1(*ALMS1*):c.1951G>T (p.Ala651Ser)	Het	0	0	VUS (PM2, BP4)	-
5166	M	GH, LH/FSH, TSH, PRL	-	Hypo	NM_016952.5(*CDON*):c.746C>T (p.Pro249Leu)	Het	0.003422	0	VUS (PM2)	-
					NM_144773.4(*PROKR2*):c.421G>A (p.Val141Ile)	Het	0.000002	0	VUS (PM2)	-
5176	F	GH, LH/FSH, TSH, ACTH	-	Hypo, EN	none	-	-	-	-	-
5179	F	GH	-	PSI, EN	NM_023110.3(*FGFR1*):c.2464C>T (p.Arg822Cys)	Het	0.000555	0.002	VUS (PP2, BP6)	18985070
					NM_000823.4(*GHRHR*):c.317C>T (p.Pro106Leu)	Het	0	0	VUS (PM2, PP3)	-
					NM_000425.5(*L1CAM*):c.1143G>C (p.Lys381Asn)	Het	0.000056	0	VUS (PM2, BP4)	-
					NM_003682.4(*MADD*):c.605G>C (p.Cys202Ser)	Het	0	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_000435.3(*NOTCH3*):c.2941C>T (p.His981Tyr)	Het	0.000160	0.004	VUS (PM2, PP2, BP6)	24086431
5188	F	GH	Neonatal hypoglycemia	EN	none	-	-	-	-	-
5197	M	GH, LH/FSH, TSH	-	Hypo, PSI	NM_000085.3(*CLCNKB*):c.490G>C (p.Gly164Arg)	Het	0.000032	0	VUS (PM2, PP3, BS2)	32203225
					NM_000142.4(*FGFR3*):c.1078G>A (p.Glu360Lys)	Het	0.000055	0	VUS (PM1, PM2)	19215249
					NM_033661.5(*WDR4*):c.1148T>C (p.Leu383Pro)	Het	0.000032	0	VUS (PM2)	-
					NM_033661.5(*WDR4*):c.1150G>A (p.Glu384Lys)	Het	0.000331	0	VUS (PM2, BP4)	-
5198	M	GH, TSH	-	N	NM_001378454.1(*ALMS1*):c.9871T>C (p.Ser3291Pro)	Het	0.000047	0	VUS (PM2, BP4)	-
					NM_001457.4(*FLNB*):c.1644_1648dupTATGC (p.Gln550fsTer25)	Het	0	0.002	LP (PVS1, PM2)	-
					NM_003482.3(*KMT2D*):c.2152C>G (p.Leu718Val)	Het	0.000004	0	VUS (PM2, PP2, BP6)	-
5199	F	GH	-	N	NM_018489.2(*ASH1L*):c.1934G>T (p.Arg645Ile)	Het	0	0	VUS (PM2, PP2)	-
					NM_015662.3(*IARS2*):c.1925C>T (p.Ala642Val)	Het	0.000177	0	VUS (PM2, BP4)	-
					NM_002834.5(*PTPN11*):c.1594G>A (p.Glu532Lys)	Het	0.000054	0	VUS (PP2)	20578946
5200	M	GH	-	N	NM_000384.2(*APOB*):c.11272G>T (p.Val3758Phe)	Het	0.000058	0	VUS (PM2, BP4)	-
5201 (a)	M	GH	Mental retardation, deafness	N	NM_030630.2(*HID1*):c.427G>A (p.Glu143Lys)	Het	0.000303	0	VUS (PM2)	-
					NM_015662.3(*IFT172*):c.4598C>T (p.Thr1533Met)	Het	0.000075	0	VUS (PM2, BP4)	-
					NM_001197104.2(*KMT2A*):c.184G>A (p.Ala62Thr)	Het	0.000065	0	VUS (PM2, PP2, BP4)	-
					NM_002941.4(*ROBO1*):c.505C>T (p.Arg169Trp)	Het	0.000004	0	VUS (PM2)	-
5202 (a)	M	GH	Mental retardation, deafness	N	NM_030630.2(*HID1*):c.427G>A (p.Glu143Lys)	Het	0.000303	0	VUS (PM2)	-
					NM_001197104.2(*KMT2A*):c.184G>A (p.Ala62Thr)	Het	0.000065	0	VUS (PM2, PP2, BP4)	-
					NM_018077.3(*RBM28*):c.1370G>A (p.Gly457Asp)	Het	0.000964	0	VUS (PM2, BS2)	-
5230	M	GH	-	N	NM_022475.3(*HHIP*):c.1762C>T (p.Pro588Ser)	Het	0.003485	0.002	VUS (PM2, BS2)	29522511
					DUP:chrX:149487173-149526099 (38.9 Kb, including part of *IDS*) [§]	-	-	-	VUS (0.30)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
5231	F	GH, TSH	Mental retardation, SOD	n/a	NM_001111035.3(*ACP5*):c.149A>G (p.Asn50Ser)	Het	0.000054	0	VUS (PM2, BP4)	-
					NM_014694.4(*ADAMTSL2*):c.2492C>T (p.Thr831Met)	Het	0.000160	0	VUS (PM2, BP4)	-
					NM_138422.4(*ADAT3*):c.854A>T (p.Asp285Val)	Het	0	0	VUS (PM2, BP4)	-
					NM_000303.3(*PMM2*):c.710C>G (p.Thr237Arg)	Het	0.000204	0.002	P (PS3, PM1, PM2, PM3, PM5, PP2, PP3)	9497260
5261	M	GH, LH/FSH, TSH	Congenital heart disease	EN	none	-	-	-	-	-
5264 (b)	F	GH	-	Hypo	NM_004321.8(*KIF1A*):c.4982T>C (p.Met1661Thr)	Het	0	0	VUS (PM2, PP2)	-
					NM_002474.3(*MYH11*):c.5767G>A (p.Ala1923Thr)	Het	0.000115	0.002	VUS (PM2)	-
					NM_004260.4(*RECQL4*):c.3212G>A (p.Arg1071His)	Het	0.000078	0	VUS (PM2)	-
5265 (b)	M	GH	-	n/a	NM_004260.4(*RECQL4*):c.3212G>A (p.Arg1071His)	Het	0.000078	0	VUS (PM2)	-
5266	M	GH	-	N	NM_001854.4(*COL11A1*):c.3650C>T (p.Pro1217Leu)	Het	0.000058	0	VUS (PM2, PP3)	-
					NM_003682.4(*MADD*):c.288C>A (p.Asn96Lys)	Het	0	0	VUS (PM2)	-
5267	M	GH	Epilepsy	N	NM_152730.6(*TBC1D32*):c.949A>G (p.Ile317Val)	Het	0.000008	0	VUS (PM2, BP4)	-
					NM_020928.2(*ZSWIM6*):c.225G>C (p.Glu75Asp)	Het	0.000001	0	VUS (PM2, PP2, BP4)	-
5269	M	GH	-	N	NM_000045.2(*ARG1*):c.22A>G (p.Ile8Val)	Het	0.000388	0	VUS (PM2, PM5, PP2)	-
					NM_030630.2(*HID1*):c.490C>T (p.Arg164Trp)	Het	0.000416	0	VUS (PM2, BS2)	-
					NM_002312.3(*LIG4*):c.2549C>T (p.Ala850Val)	Het	0.000194	0	VUS (PM2)	-
					NM_000303.3(*PMM2*):c.252A>T (p.Arg84Ser)	Het	0.000005	0	VUS (PM1, PM2, PP2)	-
5270	M	GH, LH/FSH, TSH, ACTH	SOD, mental retardation, micropenis	EN	NM_016952.5(*CDON*):c.1619G>T (p.Arg540Ile)	Het	0.000047	0	VUS (PM2, BP4)	-
					NM_015662.3(*IFT172*):c.247A>G (p.Ile83Val)	Het	0.000115	0	VUS (PM2, BP4)	-
					NM_015102.5(*NPHP4*):c.3538A>T (p.Ile1180Phe)	Het	0.000002	0.002	VUS (PM2)	-
5271	F	GH, TSH, ACTH	-	Hypo, PSI, EN	NM_004380.3(*CREBBP*):c.3233C>T (p.Ser1078Leu)	Het	0.000006	0.002	VUS (PM2, PP2)	-
5301	M	GH, LH/FSH, TSH, PRL, ACTH	-	Hypo, EN	NM_022475.3(*HHIP*):c.1577T>C (p.Val526Ala)	Het	0.000016	0.002	VUS (PM2, BP4)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
5302	M	GH, TSH	-	Hypo, EN	DUP:chr1:16035315-16057367 (22.1 Kb, including *CLCNKB*) [§]	-	-	-	VUS (-0.87)	-
5303	M	GH	-	n/a	NM_000823.4(*GHRHR*):c.1238G>A (p.Arg413His)	Het	0.000088	0	VUS (PM2, BP4)	-
					NM_004321.8(*KIF1A*):c.1063C>T (p.Arg355Cys)	Het	0.000004	0	VUS (PM1, PM2, PP2, PP3)	-
					NM_002834.5(*PTPN11*):c.417G>C (p.Glu139Asp)	**Het**	**0**	**0**	**P (PS1, PS2, PS3, PM2, PM5, PP1, PP2, PP3)**	**11992261**
5345	F	GH, LH/FSH, TSH, ACTH	Neonatal hypoglycemia	Hypo, EN	NM_000383.4(*AIRE*):c.908G>A (p.Arg303Gln)	Het	0.000141	0.002	VUS (PM2, BP6)	-
5346	F	GH	Single central incisor	ES	NM_004459.7(*BPTF*):c.7741A>T (p.Ser2581Cys)	Het	0.000011	0	VUS (PM2, PP2)	-
					NM_000085.3(*CLCNKB*):c.1938C>G (p.Asn646Lys)	Het	0.000006	0	VUS (PM2)	-
					NM_000823.4(*GHRHR*):c.1034T>C (p.Phe345Ser)	Het	0	0	VUS (PM2)	-
					NM_001197104.2(*KMT2A*):c.9379C>T (p.Pro3127Ser)	Het	0.000013	0	VUS (PM2, PP2)	-
					NM_003482.3(*KMT2D*):c.9431C>T (p.Ala3144Val)	Het	0.000014	0.002	VUS (PM2, PP2, BP6)	-
					NM_002427.4(*MMP13*):c.929G>A (p.Arg310His)	Het	0.000036	0	VUS (PM2)	-
					NM_000439.5(*PCSK1*):c.80C>T (p.Ala27Val)	Het	0.000057	0	VUS (PM2, BP4)	-
5399	M	GH	-	Hypo, EN	NM_001174116.3(*DMXL2*):c.716C>T (p.Ser239Leu)	Het	0.000002	0	VUS (PM2, PP3)	-
					NM_001457.4(*FLNB*):c.1196A>G (p.Lys399Arg)	Het	0.000857	0.007	VUS (BP6)	-
					NM_005270.5(*GLI2*):c.1445A>C (p.Lys482Thr)	Het	0.000077	0.002	VUS (PM2, PP3, BS2)	34387403
					NM_015311.2(*OBSL1*):c.4499A>G (p.Gln1500Arg)	Het	0.000006	0	VUS (PM2)	-
5400	M	GH	-	Hypo	NM_001369268.1(*ACAN*):c.8C>A (p.Thr3Asn)	Het	0	0	VUS (PM2, BP4)	-
					NM_000089.4(*COL1A2*):c.1523G>T (p.Gly508Val)	**Het**	**0**	**0**	**LP (PM1, PM2, PM5, PP2, PP3)**	**30715774**
					NM_001457.4(*FLNB*):c.5906G>A (p.Arg1969Gln)	Het	0.000022	0	VUS (PM2, BP6)	-
					NM_018124.4(*RFWD3*):c.1483A>G (p.Met495Val)	Het	0.000722	0	VUS (PM2, BP4)	-
					NM_005413.4(*SIX3*):c.318G>C (p.Glu106Asp)	Het	0.000001	0.002	VUS (PM1, PM2, PP2)	-
5401	F	GH	-	Hypo, EN	NM_015662.3(*IFT172*):c.854A>G (p.Asn285Ser)	Het	0.000001	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
5403	F	GH, LH/FSH, TSH, ACTH	-	Hypo	NM_001174116.3(*DMXL2*):c.1318C>T (p.Arg440Trp)	Het	0.000163	0.002	VUS (PM2)	-
					NM_015909.2(*NBAS*):c.3860T>C (p.Leu1287Ser)	Het	0.000066	0	VUS (PM2, BP6)	-
5426	M	GH, TSH	-	Hypo, EN	none	-	-	-	-	-
5427	M	GH	Mental retardation	N	NM_017780.4(*CHD7*):c.7535G>A (p.Arg2512Lys)	Het	0	0	VUS (PM2, PP2)	-
					NM_001126106.4(*SLC7A7*):c.605G>A (p.Gly202Asp)	Het	0.000177	0	LP (PM2, PP3)	-
5428	F	GH, TSH, ACTH, ADH	Neonatal hypoglycemia and hyponatremia	N	NM_001378454.1(*ALMS1*):c.140_151dupAGGAAGAGGCGG (p.Glu47_Ala50dup)	Het	0.000001	0	VUS (PM2, BP3)	-
5429	M	GH, TSH, ACTH	Neonatal hypoglycemia and hypotonia	Hypo, PSI, EN	none	-	-	-	-	-
5430	F	GH, TSH, ACTH	-	n/a	NM_000823.4(*GHRHR*):c.253T>C (p.Phe85Leu)	Het	0.000006	0	VUS (PM2)	-
5498	F	GH	-	N	NM_000061.3(*BTK*):c.1252T>C (p.Tyr418His)	Het	0.000354	0.007	VUS (PM2, PP2, BP6)	11472359
					NM_006767.4(*LZTR1*):c.563G>A (p.Trp188Ter)	Het	0	0	VUS (PM2)	-
					NM_000529.2(*MC2R*):c.170C>A (p.Ala57Glu)	Het	0.000429	0	VUS (PM1, PM2, BP4)	-
					NM_021224.6(*ZNF462*):c.6907G>C (p.Asp2303His)	Het	0	0	VUS (PM2, PP2, BP4)	-
5539	M	GH	-	N	NM_000135.4(*FANCA*):c.396T>G (p.Ser132Arg)	Het	0.000001	0	VUS (PM2, BP4)	-
					NM_015311.2(*OBSL1*):c.190G>A (p.Gly64Ser)	Het	0	0	VUS (PM2)	-
5602	M	GH	-	N	NM_021096.4(*CACNA1I*):c.1798G>A (p.Gly600Arg)	Het	0.000019	0	VUS (PM2, PP2)	-
					NM_001174116.3(*DMXL2*):c.1463C>T (p.Thr488Met)	Het	0.001525	0.002	LP (PS4, PM2)	-
					NM_006767.4(*LZTR1*):c.1723G>A (p.Asp575Asn)	Het	0.000541	0.002	VUS (BP4)	-
					NM_015102.5(*NPHP4*):c.3644C>T (p.Ser1215Leu)	Het	0.000005	0	VUS (PM2, BP4)	-
5650	M	GH	-	N	NM_001111035.3(*ACP5*):c.802A>G (p.Lys268Glu)	Het	0.000001	0.002	VUS (PM2, PM4)	-
					NM_016952.5(*CDON*):c.3238A>G (p.Thr1080Ala)	Het	0.000009	0	VUS (PM2, BP4)	-
					NM_015102.5(*NPHP4*):c.394A>T (p.Ile132Phe)	Het	0.000019	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_003995.3(*NPR2*):c.2543G>A (p.Arg848Gln)	Het	0.000153	0	VUS (PM2, PP2)	-
					NM_020436.3(*SALL4*):c.2990C>T (p.Thr997Ile)	Het	0	0	VUS (PM2)	-
5682	F	GH, TSH	-	Hypo, EN	NM_000088.4(*COL1A1*):c.2789C>T (p.Pro930Leu)	Het	0	0	VUS (PM2, PP2)	-
					NM_000168.6(*GLI3*):c.245G>A (p.Arg82Lys)	Het	0.000032	0.002	VUS (PM2, BP6)	27502037
					NM_019066.5(*MAGEL2*):c.169G>A (p.Ala57Thr)	Het	0.000001	0	VUS (PM2)	-
5683	M	GH	-	N	NM_000088.4(*COL1A1*):c.8G>T (p.Ser3Ile)	Het	0	0	VUS (PM2, PP2, PP3)	-
					NM_002584.3(*PAX7*):c.644G>A (p.Arg215His)	Het	0.000679	0	VUS (PM2, PP3)	-
					NM_001035256.3(*POMC*):c.706C>G (p.Arg236Gly)	Het	0.005113	0.005	VUS (PP3, BP6)	11244459
5684	M	GH	Atrial and ventricular septal defects	Hypo, PSI	**NM_005270.5(*GLI2*):c.2566dupG (p.Asp856 GlyfsTer175)**	**Het**	**0**	**0**	**LP (PVS1, PM2)**	**-**
					NM_003995.3(*NPR2*):c.472G>T (p.Gly158Trp)	Het	0	0	VUS (PM2, PP3)	-
5687	M	GH	-	N	NM_001174116.3(*DMXL2*):c.1436G>A (p.Arg479Gln)	Het	0.000041	0	VUS (PM2, BP4)	-
					NM_003482.3(*KMT2D*):c.2062C>T (p.Arg688Cys)	Het	0.000137	0.002	VUS (PM2, PP2, BP6)	-
5688	M	GH	-	N	NM_001170961.2(*IGSF1*):c.2696G>A (p.Arg899His)	Hemi	0.000260	0	VUS (PM2, BP4)	-
5689	M	GH	-	N	NM_021096.4(*CACNA1I*):c.1229G>A (p.Arg410Gln)	Het	0.000072	0	VUS (PM2, PP2)	-
					NM_002524.5(*NRAS*):c.128A>G (p.Gln43Arg)	Het	0	0	VUS (PM2, PP2, PP3)	-
5890	F	GH, TSH	-	Hypo	NM_005215.4(*DCC*):c.527A>G (p.Asn176Ser)	Het	0.000895	0.002	VUS (BS2)	24808016
					NM_012233.2(*RAB3GAP1*):c.1002T>G (p.Ile334Met)	Het	0	0	VUS (PM2, BP4)	-
5891	M	GH	-	EN	NM_018489.2(*ASH1L*):c.4136A>G (p.Tyr1379Cys)	Het	0.000001	0	VUS (PM2, PP2)	-
					NM_006767.4(*LZTR1*):c.1019G>A (p.Arg340Gln)	Het	0.000023	0	VUS (BP4)	-
					NM_004589.4(*SCO1*):c.792G>T (p.Met264Ile)	Het	0.000014	0	VUS (PM2, PP3)	-
5906	M	GH	-	N	NM_015662.3(*IFT172*):c.4542T>G (p.Cys1514Trp)	Het	0.000437	0	VUS (BS2)	-
5910	M	GH, TSH, ACTH	Neonatal hypoglycemia, micropenis	PSI	NM_019023.5(*PRMT7*):c.376G>A (p.Val126Met)	Het	0	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
5914	M	GH	Intraventricular hemorrhage, pyelocalyceal dilatation, hyaline membrane disease	Hypo	NM_000267.3(*NF1*):c.89C>T (p.Thr30Ile)	Het	0	0	VUS (PM2, PP2)	-
5939	M	GH	-	N	none	-	-	-	-	-
5986	M	GH, TSH	-	N	NM_001174116.3(*DMXL2*):c.3906_3911delATCTAA (p.Lys1302_Ser1303del)	Het	0.000001	0	VUS (PM2, PM4)	-
					NM_032195.3(*SON*):c.2024T>C (p.Val675Ala)	Het	0.000062	0	VUS (PM2, BP4)	-
6023	M	GH	Craniofacial dysmorphism, camptodactyly, coarctation of the aorta	Hypo	NM_018082.6(*POLR3B*):c.1244T>C (p.Met415Thr)	Het	0.001099	0.002	VUS (PP2, PP3, PP5, BS1, BS2)	25133958
					NM_144773.4(*PROKR2*):c.1000G>A (p.Val334Met)	Het	0.000036	0	VUS (PM2, PP3)	24031091
6025	M	GH	-	Hypo	NM_002633.3(*PGM1*):c.1411G>A (p.Ala471Thr)	Het	0.000011	0	VUS (PM2)	-
					NM_019023.5(*PRMT7*):c.1093G>T (p.Val365Leu)	Het	0	0	VUS (PM2, BP4)	-
6128	F	GH, TSH	-	Hypo	NM_002584.3(*PAX7*):c.335C>T (p.Pro112Leu)	Het	0.002998	0	VUS (PM2, PP3, BS2, BP6)	-
6129	F	GH	-	Hypo	DUP:chr16:14822793-16371662 (1548.9 Kb, including *MYH11*) [§]	-	-	-	VUS (0.75)	-
					NM_015093.6(*TAB2*):c.1730T>C (p.Leu577Pro)	Het	0.000013	0	VUS (PM2)	-
6180	M	GH, LH/FSH, TSH, ACTH	Neontal hypoglycemia, cryptorchidism, micropenis	Hypo, PSI, EN	NM_003682.4(*MADD*):c.4307G>A (p.Arg1436Gln)	Het	0.001528	0	VUS (PM2, BS2)	38459224
					NM_015102.5(*NPHP4*):c.593C>T (p.Ala198Val)	Het	0.000058	0.002	VUS (PM2)	-
					NM_003995.3(*NPR2*):c.2644G>A (p.Val882Ile)	Het	0.000207	0	VUS (PM2, PP2, PP3)	-
					NM_015311.2(*OBSL1*):c.4322G>A (p.Arg1441Gln)	Het	0.000059	0.002	VUS (PM2, BP4)	-
					NM_000439.5(*PCSK1*):c.541T>C (p.Tyr181His)	Het	0.000296	0	LP (PM2, PP3)	22210313
6181	F	GH, TSH, ACTH, ADH	Neonatal hypoglycemia and cholestatic jaundice	Hypo	NM_005215.4(*DCC*):c.4001C>G (p.Ala1334Gly)	Het	0	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
6182	M	GH, LH/FSH, TSH, ACTH, ADH	SOD, mental retardation, deafness, micropenis	n/a	NM_017780.4(*CHD7*):c.4895G>A (p.Arg1632His)	Het	0.000004	0	VUS (PM2, PP2, BP6)	-
					NM_015102.5(*NPHP4*):c.3644C>T (p.Ser1215Leu)	Het	0.000005	0	VUS (PM2, BP4)	-
					NM_144773.4(*PROKR2*):c.868C>T (p.Pro290Ser)	**Het**	**0.000260**	**0**	**P (PS3, PS4, PM2, PP3)**	**17054399**
6183	M	GH	-	Hypo	NM_015311.2(*OBSL1*):c.4484G>A (p.Arg1495His)	Het	0.000402	0	VUS	-
					NM_018117.12(*TCF7L1*):c.1570T>C (p.Ser524Pro)	Het	0.000001	0	VUS (PM2)	-
6184	M	GH	-	Hypo, EN	NM_021096.4(*CACNA1I*):c.3691G>A (p.Gly1231Ser)	Het	0.000183	0	VUS (PM2, PP2, PP3, BP6)	-
					NM_000168.6(*GLI3*):c.2587C>T (p.Arg863Cys)	Het	0.000018	0	VUS (PM2, PP3)	-
					NM_002755.4(*MAP2K1*):c.1180dupT (p.Ter394LeuextTer112)	Het	0	0	VUS (PM2, PM4)	-
6238	M	GH	-	Hypo, EN	NM_000057.4(*BLM*):c.3164G>C (p.Cys1055Ser)	Het	0.000007	0	P (PS3, PM2, PM3, PM5, PP3)	7585968
					NM_005215.4(*DCC*):c.4027C>T (p.Arg1343Cys)	Het	0.000265	0.002	VUS (PM2)	-
					NM_015909.2(*NBAS*):c.4916T>C (p.Leu1639Pro)	Het	0.000001	0	VUS (PM2, PP3)	-
6239	M	GH	SOD	n/a	DUP:chr1:16039531-16060002 (20.5 Kb, including *CLCNKB*) [§]	-	-	-	VUS (-0.87)	-
6240	F	GH, LH/FSH, TSH, ACTH	-	ES, Hypo	none	-	-	-	-	-
6259	M	GH	-	Hypo, EN	NM_006260.5(*DNAJC3*):c.206A>G (p.Asp69Gly)	Het	0.000665	0.002	VUS (PM2)	-
					NM_015311.2(*OBSL1*):c.4484G>A (p.Arg1495His)	Het	0.000402	0	VUS	-
					NM_006702.5(*PNPLA6*):c.3827A>G (p.Tyr1276Cys)	Het	0	0	VUS (PM2, PP2)	-
6260	M	GH	-	Hypo	NM_001378454.1(*ALMS1*): c.196G>T (p.Glu66Ter)	Het	0	0	P (PVS1, PM2, PP5)	-
					NM_000045.2(*ARG1*):c.878T>C (p.Val293Ala)	Het	0	0	VUS (PM2, PP2)	-
					NM_001374820.1(*ARID1B*):c.6255C>A (p.Asp2085Glu)	Het	0	0	VUS (PM2, BP4)	-
					NM_001851.4(*COL9A1*):c.1150C>T (p.Pro384Ser)	Het	0.000033	0	VUS (PM2, BP1)	-
					NM_006766.5(*KAT6A*):c.5299C>G (p.His1767Asp)	Het	0	0	VUS (PM2, PP3)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_005994.4(*TBX2*):c.953C>A (p.Ala318Glu)	Het	0.000171	0	VUS (PM2)	-
6262	M	GH, TSH, ACTH	Neonatal hypoglycemia, vesicoureteral reflux	PSI, EN	NM_000384.2(*APOB*):c.2677G>C (p.Asp893His)	Het	0	0	VUS (PM2)	-
6292	F	GH	-	N	NM_001369268.1(*ACAN*):c.4639G>A (p.Gly1547Arg)	Het	0.000542	0	VUS (PM2)	-
					NM_000135.4(*FANCA*):c.2467A>T (p.Thr823Ser)	Het	0.000001	0	VUS (PM2, BP4)	-
					NM_003482.3(*KMT2D*):c.80G>A (p.Ser27Asn)	Het	0.000029	0	VUS (PM2, PP2, BP6)	-
					NM_021224.6(*ZNF462*):c.1997C>T (p.Ser666Phe)	Het	0.000005	0	VUS (PM2, PP2)	-
6293	M	GH	-	Hypo	NM_001174116.3(*DMXL2*):c.5974G>A (p.Asp1992Asn)	Het	0.0002514	0	VUS (PM2, BP4)	-
					NM_003482.3(*KMT2D*):c.1028A>G (p.Tyr343Cys)	Het	0.000001	0	VUS (PM2, PP2)	-
					NM_003482.3(*KMT2D*):c.14957G>A (p.Arg4986His)	Het	0.000004	0	VUS (PM2, PP2, BP6)	-
					NM_002474.3(*MYH11*):c.1153A>C (p.Met385Leu)	Het	0	0	VUS (PM2, BP4)	-
6294	M	GH, TSH	Microcephaly, mental retardation, mild facial dysmorphism, cryptorchidism	Hypo, EN	NM_001457.4(*FLNB*):c.4396G>A (p.Val1466Met)	Het	0.000010	0	VUS (PM2)	-
					NM_001197104.2(*KMT2A*):c.9379C>T (p.Pro3127Ser)	Het	0.000013	0	VUS (PM2, PP2)	-
					NM_000435.3(*NOTCH3*):c.2932A>C (p.Ser978Arg)	Het	0.000447	0.002	VUS (PM2, PP2, BP6)	17879447
					NM_015102.5(*NPHP4*):c.517C>T (p.Gln173 Ter)	Het	0.000004	0	P (PVS1, PM2, PM3)	23559409
					DUP:chrX:135718920-149769070 (14050.2 Kb, including *SOX3* and *IDS*) [§]	**-**	**-**	**-**	**P (1.00)**	**-**
6318	M	GH, TSH, ACTH	Neonatal hypoglycemia, epileptic encephalopathy, SOD, mental retardation	Hypo, EN	NM_014112.5(*TRPS1*):c.3574G>A (p.Gly1192Ser)	Het	0.000072	0	VUS (PM2, PP3, BP6)	-
6350	F	GH	-	N	NM_001369268.1(*ACAN*):c.229C>T (p.Arg77Cys)	Het	0.000035	0	VUS (PM2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
					NM_001202.6(*BMP4*):c.734G>A (p.Arg245Gln)	Het	0.000020	0.002	VUS (PM2, BP4)	-
					NM_001197104.2(*KMT2A*):c.568C>T (p.Leu190Phe)	Het	0	0	VUS (PM2, PP2)	-
6383	M	GH	-	EN	NM_138387.4(*G6PC3*):c.772G>T (p.Ala258Ser)	Het	0	0	VUS (PM1, PM2)	-
6486	M	GH, TSH, ACTH	Neonatal hypoglycemia	EN	NM_000383.4(*AIRE*):c.708C>G (p.Phe236Leu)	Het	0	0	VUS (PM2)	-
					NM_015311.2(*OBSL1*):c.3117G>T (p.Arg1039Ser)	Het	0.000288	0	VUS (PM2, BP4)	-
					NM_018344.6(*SLC29A3*):c.988A>G (p.Ile330Val)	Het	0.000476	0	VUS (BS2)	-
6519	F	GH, TSH	-	Hypo, EN	NM_000515.5(*GH1*):c.517G>C (p.Asp173His)	Het	0	0	VUS (PM2)	-
					DEL:chr2:117814697-121765380 (3950.7 Kb, including *GLI2*) [§]	**-**	**-**	**-**	**P (1.00)**	**-**
6543	M	GH, TSH, ACTH	Neonatal hypoglycemia, ocular malformations	Hypo, EN	NM_000142.4(*FGFR3*):c.1201C>G (p.Pro401Ala)	Het	0.000128	0.005	VUS (PM2)	-
6775	F	GH, TSH, ACTH	-	Hypo, EN	NM_016952.5(*CDON*):c.1982C>A (p.Ala661Glu)	Het	0.000001	0	VUS (PM2)	-
					NM_017780.4(*CHD7*):c.1554G>T (p.Gln518His)	Het	0.000018	0.002	VUS (PM2, PP2, BP6)	-
					NM_015311.2(*OBSL1*):c.2530G>A (p.Gly844Arg)	Het	0.000026	0	VUS (PM2)	-
					NM_012233.2(*RAB3GAP1*):c.244G>A (p.Glu82Lys)	Het	0	0.002	VUS (PM2, BP4)	-
6800	M	GH	-	N	NM_006766.5(*KAT6A*):c.3862G>A (p.Glu1288Lys)	Het	0.000166	0	VUS (PM2, BP6)	-
					NM_001197104.2(*KMT2A*):c.10591C>G (p.Pro3531Ala)	Het	0.000077	0	VUS (PM2, PP2, BP6)	-
					NM_000439.5(*PCSK1*):c.1168G>A (p.Gly390Ser)	Het	0.000066	0	LP (PM2, PP3)	-
6868	F	GH, LH/FSH, TSH, PRL	-	N	NM_016952.5(*CDON*):c.2036C>T (p.Ala679Val)	Het	0.000255	0	VUS (PM2)	-
					NM_015311.2(*OBSL1*):c.1039C>A (p.Leu347Met)	Het	0.0000003	0	VUS (PM2)	-
					NM_007373.4(*SHOC2*):c.1660A>G (p.Ser554Gly)	Het	0.000090	0	VUS (PP2)	-
6903	F	GH	-	N	NM_006766.5(*KAT6A*):c.3598C>T (p.Arg1200Cys)	Het	0.000023	0	VUS (PM2, BP6)	-
6910	M	GH, LH/FSH, TSH, ACTH	Pectus excavatum, scoliosis, facial dysmorphism,	Hypo, EN	**DEL:chr14:52671585-56730336 (4058.8 Kb, including *BMP4*)** [§]	**Het**	**-**	**-**	**P (1.00)**	**-**

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
			cataracts, blue sclera							
					NM_002181.3(*IHH*):c.877A>G (p.Thr293Ala)	Het	0.000009	0	VUS (PM2, PP2)	-
					NM_000267.3(*NF1*):c.3436G>A (p.Val1146Ile)	**Het**	**0.000226**	**0**	**LP (PM1, PM2, PM5, PP2, BP6)**	**Doi:10.1590/S1415-47572004000300003**
					NM_002524.5(*NRAS*):c.554C>T (p.Pro185Leu)	Het	0.000007	0	VUS (PP2, BP4)	-
6924	M	GH	-	Hypo	none	-	-	-	-	-
6930	M	GH, LH/FSH, TSH, ACTH	-	n/a	NM_000135.4(*FANCA*):c.1340C>T (p.Ser447Leu)	Het	0.000329	0	VUS (PM1, PM2)	26492932
					NM_023110.3(*FGFR1*):c.584A>G (p.Lys195Arg)	Het	0.000032	0.002	VUS (PM2, PP2, BP4)	-
					NM_006766.5(*KAT6A*):c.4778C>T (p.Ser1593Leu)	Het	0.000011	0	VUS (PM2, BP6)	-
					NM_014564.5(*LHX3*):c.43G>A (p.Gly15Arg)	Het	0	0	VUS (PM2)	-
					NM_019066.5(*MAGEL2*):c.2176A>G (p.Ile726Val)	Het	0.000516	0	VUS	-
6933	F	GH, LH/FSH, TSH, ACTH	-	PSI, EN	NM_004380.3(*CREBBP*):c.6521T>C (p.Met2174Thr)	Het	0	0	VUS (PM2, PP2)	-
6949	M	GH	SOD	Hypo	NM_198407.2(*GHSR*):c.647T>C (p.Val216Ala)	Het	0.000225	0	VUS (PM2)	-
					NM_015662.3(*IFT172*):c.4564G>A (p.Glu1522Lys)	Het	0.000194	0	VUS (PM2, BP4)	-
					NM_024426.6(*WT1*):c.1063T>C (p.Cys355Arg)	Het	0.000918	0	VUS (PM5, PP2, PP3, BP6)	22703879
7017	M	GH	-	Hypo	NM_000439.5(*PCSK1*):c.1168G>A (p.Gly390Ser)	Het	0.000066	0	LP (PM2, PP3)	-
7020	M	GH, ACTH	-	Hypo, EN	NM_017780.4(*CHD7*):c.3299G>A (p.Arg1100His)	Het	0.000115	0	VUS (PM2, PP2, PP3, BP6)	31726455
					NM_015662.3(*IFT172*):c.622A>G (p.Asn208Asp)	Het	0	0	VUS (PM2, BP4)	-
7021	M	GH, TSH, ACTH	Neonatal hypoglycemia, SOD, micropenis	PSI, EN	NM_019023.5(*PRMT7*):c.2055C>G (p.Phe685Leu)	Het	0.000180	0	VUS (PM2)	-
7023	F	GH	-	N	NM_015662.3(*IFT172*):c.5053C>T (p.Pro1685Ser)	Het	0.000055	0	VUS (PM2)	-
					NM_003682.4(*MADD*):c.647G>A (p.Arg216Gln)	Het	0.000004	0	VUS (PM2, BP4)	-
					NM_002834.5(*PTPN11*):c.1678C>T (p.Leu560Phe)	Het	0.000115	0.002	VUS (PP2)	12960218
					NM_002941.4(*ROBO1*):c.4565C>T (p.Ser1522Leu)	Het	0.000990	0	VUS (PP5, BS1, BS2, BP4)	35348658

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
7024	M	GH, TSH, ACTH	Neonatal hypoglycemia, jaundice, cryptorchidism	Hypo	none	-	-	-	-	-
7025	M	GH	-	N	NM_014564.5(*LHX3*):c.811A>T (p.Met271Leu)	Het	0	0	VUS (PM2)	-
					NM_006445.4(*PRPF8*):c.5164A>G (p.Ser1722Gly)	Het	0.000225	0	VUS (PM2, PP2)	-
					NM_001126106.4(*SLC7A7*):c.954A>T (p.Leu318Phe)	Het	0.000317	0.002	VUS (PM2)	-
7026	F	GH, LH/FSH, TSH, PRL, ACTH	Neonatal hypoglycemia, hypotonia, jaundice	Hypo	NM_001126106.4(*SLC7A7*):c.1382_1384delTCA (p.Ile461del)	Het	0.000299	0	VUS (PM2, PM4)	-
7027	F	GH, TSH, ACTH	SOD	Hypo, EN	NM_000384.2(*APOB*):c.11777A>G (p.Tyr3926Cys)	Het	0.000006	0	VUS (PM2)	-
7028	M	GH	-	N	NM_005270.5(*GLI2*):c.2794C>T (p.Pro932Ser)	Het	0.000004	0	VUS (PM2)	17096318
					NM_006766.5(*KAT6A*):c.2746A>G (p.Ser916Gly)	Het	0	0	VUS (PM2, BP4)	-
					NM_002427.4(*MMP13*):c.403G>A (p.Glu135Lys)	Het	0.000032	0	VUS (PM2, BP4)	-
7032	M	GH, ACTH	Neonatal hypoglycemia and jaundice	Hypo	none	-	-	-	-	-
7033	M	GH	-	N	NM_001854.4(*COL11A1*):c.1522A>G (p.Thr508Ala)	Het	0.000374	0	VUS (BP6)	-
					NM_015662.3(*IFT172*):c.3435T>G (p.His1145Gln)	Het	0.002593	0	VUS (PM2, BS2)	-
7066	F	GH, ACTH	-	Hypo, PSI, EN	NM_018077.3(*RBM28*):c.1007C>T (p.Thr336Ile)	Het	0.000451	0	VUS (PM2, PP3)	-
7070	M	GH, TSH, ACTH	Neonatal hypoglycemia and jaundice	EN	NM_000435.3(*NOTCH3*):c.2932A>C (p.Ser978Arg)	Het	0.000447	0.002	VUS (PM2, PP2, BP6)	17879447
7073	F	GH, TSH, ACTH	-	Hypo, PSI, EN	NM_000045.2(*ARG1*):c.863_865delAAG (p.Glu288del)	Het	0.000013	0	LP (PM2, PM3, PM4)	-
					NM_014862.4(*ARNT2*):c.28C>T (p.Pro10Ser)	Het	0.000445	0.002	VUS (PM2, BP4)	-
					NM_004380.3(*CREBBP*):c.6376G>A (p.Gly2126Ser)	Het	0.000005	0	VUS (PM2, PP2)	-
					NM_000527.4(*LDLR*):c.2575G>A (p.Val859Met)	Het	0.000033	0.002	VUS (PM2, BP4)	17765246
					NM_002427.4(*MMP13*):c.301G>T (p.Val101Leu)	Het	0.000193	0	VUS (PM1, PM2, BP4)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
7075	M	GH, LH/FSH, ACTH	Anosmia	N	NM_198407.2(*GHSR*):c.1065T>A (p.Ser355Arg)	Het	0.000054	0	VUS (PM2, BP4)	-
					NM_001126128.2(*PROK2*):c.163delA (p.Ile55Ter)	**Homo**	**0.000268**	**0**	**P (PVS1, PS4, PM2)**	**17959774**
					NM_031283.3(*SHH*):c.967G>A (p.Gly323Arg)	Het	0.0000003	0	VUS (PM2, PP2)	-
7076	F	GH	-	N	DEL:chrX:118675949-135547267 (16871.3 Kb, including *IGSF1* and *PHF6*) §	Het	-	-	P (1.00)	-
					NM_014112.5(*TRPS1*):c.1417G>A (p.Gly473Arg)	Het	0.000053	0	VUS (PM2, BP6)	-
7084	F	GH	SOD, mental retardation, uterine agenesis	ES, EN	NM_001378454.1(*ALMS1*):c.2683_2685delGAT (p.Asp895del)	Het	0.000036	0	VUS (PM2, PM4)	-
					NM_001457.4(*FLNB*):c.6302G>A (p.Arg2101His)	Het	0.000165	0	VUS (PM2, BP6)	-
					NM_001170961.2(*IGSF1*):c.2807C>T (p.Thr936Ile)	Het	0.000001	0.002	VUS (PM2, BP4)	-
					NM_001077494.3(*NFKB2*):c.1214C>T (p.Ala405Val)	Het	0	0	VUS (PM2, PP2, BP4)	-
					NM_015311.2(*OBSL1*):c.4517G>A (p.Arg1506His)	Het	0.000054	0	VUS (PM2)	-
7085	M	GH	-	Hypo	NM_001374820.1(*ARID1B*):c.5438A>G (p.Lys1813Arg)	Het	0	0	VUS (PM2, BP4)	-
					NM_001851.4(*COL9A1*):c.1201A>G (p.Thr401Ala)	Het	0.000049	0	VUS (PM2, BP1)	-
					DUP:chr22:18908635-21060572 (2151.9 Kb, including *LZTR1*) §	-	-	-	P (1.00)	-
					NM_015102.5(*NPHP4*):c.2830G>A (p.Val944Ile)	Het	0.000154	0.004	VUS (PM2, BP4)	-
					NM_032195.3(*SON*):c.961A>C (p.Ser321Arg)	Het	0.000001	0	VUS (PM2, BP4)	-
					NM_153704.6(*TMEM67*):c.1700A>G (p.Tyr567Cys)	Het	0.000430	0	VUS (PM2, PP2, PP3)	22277662
7086	M	GH	-	N	NM_000089.4(*COL1A2*):c.3313G>A (p.Gly1105Ser)	Het	0.002548	0.002	VUS (PP2, PP3, PP5, BS1, BS2, BP6)	21667357
					NM_023110.3(*FGFR1*):c.566G>A (p.Arg189His)	Het	0.000043	0.002	VUS (PM2, PP2)	-
					NM_021964.3(*ZNF148*):c.683G>A (p.Arg228His)	Het	0.000043	0	VUS (PM2, PP2, BP4)	-
7087	M	GH	-	Hypo	**NM_144773.4(*PROKR2*):c.868C>T (p.Pro290Ser)**	**Het**	**0.000260**	**0**	**P (PS3, PS4, PM2, PP3)**	**17054399**
					NM_005996.4(*TBX3*):c.1450G>A (p.Ala484Thr)	Het	0.000139	0	VUS (PM2)	-
					NM_005996.4(*TBX3*):c.1451C>A (p.Ala484Asp)	Het	0.000139	0	VUS (PM2)	-
					NM_021224.6(*ZNF462*):c.3181T>C (p.Phe1061Leu)	Het	0	0	VUS (PM2, PP2)	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
7088	M	GH	-	EN	NM_016952.5(*CDON*):c.302A>G (p.Asn101Ser)	Het	0.000015	0	VUS (PM2, BP4)	-
7336	M	GH, LH/FSH	-	Hypo	NM_000383.4(*AIRE*):c.1435G>A (p.Val479Met)	Het	0.000084	0	VUS (PM2)	-
					NM_001378454.1(*ALMS1*):c.5017A>G (p.Ile1673Val)	Het	0.000004	0	VUS (PM2, BP4)	-
					NM_000425.5(*L1CAM*):c.2552C>T (p.Thr851Met)	Hemi	0.000452	0	VUS (BP6)	-
					NM_018117.12(*WDR11*):c.3571G>A (p.Gly1191Ser)	Het	0.000540	0.007	VUS (PM2, PP3, BS2)	33270637
7349	M	GH, LH/FSH	Anosmia	N	NM_002292.3(*LAMB2*):c.1762C>G (p.Pro588Ala)	Het	0.000720	0	VUS (BP4)	-
					NM_002834.5(*PTPN11*):c.1678C>T (p.Leu560Phe)	Het	0.000115	0.002	VUS (PP2)	12960218
					NM_018124.4(*RFWD3*):c.2059G>A (p.Val687Ile)	Het	0.000004	0	VUS (PM2, BP4)	-
7350	M	GH, TSH, ACTH, ADH	Neonatal hypoglycemia and hypernatremia, SOD	N	NM_001374820.1(*ARID1B*):c.2750G>A (p.Ser917Asn)	Het	0.000006	0	VUS (PM2, BP4)	-
					NM_001174116.3(*DMXL2*):c.8203G>A (p.Ala2735Thr)	Het	0.000023	0	VUS (PM2, BP4)	-
					NM_001130823.3(*DNMT1*):c.3532G>C (p.Asp1178His)	Het	0.000001	0	VUS (PM2, PP2, BP4)	-
					NM_002303.6(*LEPR*):c.1246C>T (p.His416Tyr)	Het	0.000718	0.002	VUS (PM2, BP6)	-
					NM_006767.4(*LZTR1*):c.1723G>A (p.Asp575Asn)	Het	0.000541	0.002	VUS (BP4)	-
					NM_000435.3(*NOTCH3*):c.5992C>T (p.Arg1998Cys)	Het	0.001219	0.002	VUS (PP2, BP6)	-
					NM_004260.4(*RECQL4*):c.770G>A (p.Ser257Asn)	Het	0	0	VUS (PM2)	-
					NM_007373.4(*SHOC2*):c.242C>T (p.Thr81Ile)	Het	0	0	VUS (PP2)	-
					NM_021224.6(*ZNF462*):c.5254G>A (p.Asp1752Asn)	Het	0.000895	0	VUS (PP2, BP6)	-
7356	M	GH, TSH	-	EN	NM_001111035.3(*ACP5*):c.838C>T (p.Arg280Cys)	Het	0.000011	0	VUS (PM2)	-
					NM_005270.5(*GLI2*):c.3529A>G (p.Lys1177Glu)	Het	0.000006	0	VUS (PM2, BP4)	-
7365	M	GH, LH/FSH, TSH, ACTH	Neonatal hypoglycemia, micropenis, cryptorchidism	Hypo, EN	none	-	-	-	-	-

Patient id	Sex	Pituitary Hormone Deficiencies	Other associated features	MRI	Variant (GRCh38)	Zygosity	Max Allele frequency in GnomAD	Allele frequency in Portuguese controls	ACMG/ClinGen classification (criteria or score)	First report (PMID)
7380	M	GH, LH/FSH, TSH, ACTH	-	ES	NM_015311.2(*OBSL1*):c.727C>G (p.Pro243Ala)	Het	0.000043	0	VUS (PM2, BP4)	-
7416	F	GH, TSH	Neonatal hypoglycemia	Hypo, EN	NM_000135.4(*FANCA*):c.3430C>T (p.Arg1144Trp)	Het	0.000949	0	VUS (PM2, BS2)	14695169
7462	M	GH, LH/FSH, TSH, ACTH	-	Hypo, EN	NM_032195.3(*SON*):c.4328C>T (p.Ser1443Leu)	Het	0.000058	0.004	VUS (PM2, BP4)	-
7494	M	GH, LH/FSH, TSH, ACTH	Neonatal hypoglycemia and hyponatremia, micropenis	ES, PSI	**NM_005270.5(*GLI2*):c.1330T>G (p.Cys444Gly)**	**Het**	**0**	**0**	**LP (PM2, PP3)**	-
					NM_006702.5(*PNPLA6*):c.3598C>G (p.Gln1200Glu)	Het	0.001912	0.002	VUS (PM2, PP2)	25133958
					NM_019023.5(*PRMT7*):c.2032A>G (p.Thr678Ala)	Het	0.000180	0	VUS (PM2, BP4)	-
7589	M	GH	-	N	none	-	-	-	-	-
7639	M	GH, LH/FSH, TSH	Mental retardation	Hypo, EN	NM_001378454.1(*ALMS1*):c.1237+2T>C	Het	0.000001	0.002	P (PVS1, PM2, PM3)	-
					NM_001202.6(*BMP4*):c.749A>G (p.Gln250Arg)	Het	0	0	VUS (PM2)	-
					NM_016952.5(*CDON*):c.1619G>T (p.Arg540Ile)	Het	0.000047	0	VUS (PM2, BP4)	-
					NM_015909.2(*NBAS*):c.5185T>G (p.Phe1729Val)	Het	0.000518	0	VUS (PM2)	-
					NM_006261.5(*PROP1*):c.296G>A (p.Arg99Gln)	Het	0.000066	0	P (PS3, PM2, PM3, PP1, PP3)	12519826
8229	F	GH, ADH	-	Hypo, EN	NM_001457.4(*FLNB*):c.1409G>A (p.Arg470Gln)	Het	0.000043	0	VUS (PM2, BP6)	-
8293	M	GH, LH/FSH, TSH, ACTH, PRL	Abnormal neck rotation	N	NM_025139.6(*ARMC9*):c.1865C>T (p.Thr622Met)	Het	0.000018	0	VUS (PM2)	-
					NM_001854.4(*COL11A1*):c.1023A>C (p.Glu341Asp)	Het	0	0	VUS (PM2)	-
					NM_001174116.3(*DMXL2*):c.7585A>G (p.Ile2529Val)	Het	0.000430	0.004	VUS (PM2, BP4)	-
					NM_004260.4(*RECQL4*):c.3317G>A (p.Arg1106His)	Het	0.001048	0.002	VUS (BP6)	-

id, identification code; (a-b), Familial cases; M, Male; F, Female; GH, Growth Hormone; LH, Luteinizing Hormone; FSH, Follicle–Stimulating Hormone; TSH, Thyroid Stimulating Hormone; ACTH, Adrenocorticotropic Hormone; PRL, Prolactin; ADH, Antidiuretic hormone; SOD, Septo–Optic Dysplasia; MRI, Magnetic Resonance Imaging; Hypo, Pituitary Hypoplasia; PSI, Pituitary Stalk Interruption; EN, Ectopic Neurohypophysis; ES, Empty Sella; N, Normal; RC, Rathke's Cyst; n/a, not available; GRCh38, Genome Research Consortium human build 38; in **bold**, disease-causing variants also presented in Table 1**;** DEL, deletion; Kb, kilobases; §, approximate breakpoints; DUP, duplication; Het, heterozygous; Hemi, hemizygous; Homo, homozygous; Max, Maximum; GnomAD, Genome Aggregation Database (v4.1.0); ACMG, American College of Medical Genetics

and Genomics; ClinGen, Clinical Genome Resource. Variants were classified as Pathogenic (P), Likely Pathogenic (LP), Variants of Uncertain Significance (VUS), Likely Benign (LB), or Benign (B). Sequence variants were classified based on the evidence for pathogenicity [very strong (PVS1), moderate (PM1–6), or supporting (PP1–5)] or benign impact [stand–alone (BA), strong (BS1–4), or supporting (BP1–7)]. Copy number variants were classified based on an evidence-based semi-quantitative scoring. Gene-specific ClinGen guidelines and recommendations were considered when available. PMID, PubMed identifier; DOI, Digital Object Identifier.