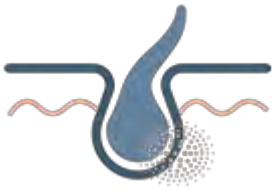
Ichthyosis

Precision Panel



Overview

Ichthyosis refers to an uncommon group of skin disorders characterized by excessive amounts of dry surface scales and hyperkeratosis, often associated with erythroderma. It belongs to the group of disorders of keratinization and the manifestations are due to mutations in genes mostly involved in skin barrier formation. Ichthyoses can be syndromic or non-syndromic. The ichthyosiform dermatoses can be classified according to clinical manifestations, genetic presentation and histologic findings which results in a clinical heterogeneity. There are five types of inherited ichthyosis as follows: ichthyosis vulgaris, lamellar ichthyosis, epidermolytic hyperkeratosis, congenital ichthyosiform erythroderma and X-linked ichthyosis.

The Igenomix Ichthyosis Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of skin scaling ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Ichthyosis Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Fine scales and varying degrees of dryness of the skin localized over the trunk, abdomen, buttocks and legs
- Conjunctiva thickening
- Keratitis, megalocornea and other corneal manifestations
- Eyelid manifestations: ectropion, blepharitis, trichiasis
- Retinal symptoms: Coloboma, tortuous vessels etc

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

- Early initiation of treatment with a multidisciplinary team in the form of medical care with oral retinoids, prevention of complications such as infections, eye care and surgical care if needed.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA12	Ichthyosis Congenita, Lamellar Ichthyosis, Congenital Non-bullous Ichthyosiform Erythroderma	AR	100	151 of 153
ALDH3A2	Sjogren-Larsson Syndrome	AR	96	119 of 119
ALOX12B	Congenital Nonbullous Ichthyosiform Erythroderma, Lamellar Ichthyosis	AR	100	80 of 80
ALOXE3	Congenital Nonbullous Ichthyosiform Erythroderma, Lamellar Ichthyosis	AR	100	27 of 27
ANOS1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	X,XR,G	96.86	NA of NA
AP1B1	Ichthyosiform Erythroderma, Corneal Involvement, And Deafness, Mednik Syndrome	AR	100	5 of 5
AP1S1	Mental Retardation, Enteropathy, Deafness, Peripheral Neuropathy, Ichthyosis, And Keratoderma, Mednik Syndrome	AR	99.98	2 of 2
ARSL	X-linked Recessive Chondrodysplasia Punctata	X,XR,G	99.99	NA of NA
ASPRV1	Ausotomal Dominant Ichthyosis Lamellar	AD	98.23	4 of 4
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Noonan Syndrome With Multiple Lentigines	AD	100	80 of 80
CARD14	Pityriasis Rubra Pilaris, Psoriasis	AD	99.95	47 of 47
CARMIL2	Immunodeficiency	AR	96.16	NA of NA
CASP14	Autosomal Recessive Congenital Ichthyosis	AR	100	1 of 1
CCDC141	Hypogonadotropic Hypogonadism Without Anosmia, Kallmann Syndrome	AR	99.7	1 of 1
CDSN	Hypotrichosis, Peeling Skin Syndrome	AD,AR	99.88	12 of 13
CERS3	Autosomal Recessive Congenital Ichthyosis, Congenital Non-Bullous Ichthyosiform Erythroderma	AR	99.98	9 of 9
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism With Or Without Anosmia, Omenn Syndrome	AD	96.25	823 of 896
CHKB	Congenital Muscular Dystrophy, Megaconial Type	AR,MI	100	29 of 29
CLDN1	Ichthyosis, Leukocyte Vacuoles, Alopecia, And Sclerosing Cholangitis, Neonatal Ichthyosis-Sclerosing Cholangitis Syndrome	AR	100	4 of 4
COL4A5	Alport Syndrome	X,XD,G	99.88	NA of NA
CSTA	Autosomal Recessive Exfoliative Ichthyosis, Ichthyosis Bullosa Ofsiemens-like, Acral Peeling Skin Syndrome	AR	100	5 of 5
CYP4F22	Autosomal Recessive Congenital Ichthyosis, Lamellar Ichthyosis	AR	100	50 of 50

DCC	Gaze Palsy, Familial Horizontal, With Progressive Scoliosis With Impaired Intellectual Development, Familial Congenital Mirror Movements	AD,AR	94	39 of 39
DNAJC21	Bone Marrow Failure Syndrome, Shwachman-Diamond Syndrome	AR	99.83	12 of 12
DOLK	Congenital Disorder Of Glycosylation Type Im	AR	99.98	13 of 13
DSP	Lethal Acantholytic Epidermolysis Bullosa, Keratosis Palmoplantaris Striata II, Skin Fragility-Woolly Hair Syndrome, Carvajal Syndrome	AD,AR	99.91	366 of 369
DUSP6	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.36	4 of 4
EBP	X-linked Dominant Chondrodysplasia Punctata, Mend Syndrome	X,XR,XD,G	100	NA of NA
EFL1	Shwachman-Diamond Syndrome	AR	99.94	NA of NA
ELOVL4	Erythrokeratodermia With Ataxia, Ichthyosis, Spastic Quadriplegia, And Mental Retardation, Stargardt Disease, Spinocerebellar Ataxia Type 34	AD,AR	100	16 of 17
EMD	X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92	NA of NA
ERCC2	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum Complementation Group D, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
ERCC3	Photosensitive Trichothiodystrophy, Xeroderma Pigmentosum Complementation Group B, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.98	24 of 24
FEZF1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AR	99.95	3 of 3
FGF17	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.98	8 of 8
FGF8	Hypogonadotropic Hypogonadism With Or Without Anosmia, Holoprosencephaly, Kallmann Syndrome	AD	98.36	38 of 38
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Osteoglophonic Dysplasia, Pfeiffer Syndromes, Septo-Optic Dysplasia Spectrum	AD	100	279 of 280
FHL1	X-linked Reducing Body Myopathy, X-linked Dominant Scapuloperoneal Myopathy, Uruguay Faciocardiomusculoskeletal Syndrome, X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,XD,G	99.98	NA of NA
FITM2	Siddiqi Syndrome	AR	99.97	3 of 3
FLG	Ichthyosis Vulgaris	AD	98.03	111 of 119
FLRT3	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD	99.98	7 of 7
GBA	Fetal Gaucher Disease , Gaucher Disease-Ophthalmoplegia-Cardiovascular Calcification Syndrome	AD,AR	100	469 of 471
GINS1	Immunodeficiency	AR	99.87	5 of 5
GJA1	Alopecia Congenita With Keratosis Palmoplantaris, Autosomal Recessive Craniometaphyseal Dysplasia, Erythrokeratodermia Variabilis Et Progressiva, Oculodentodigital Dysplasia	AD,AR,MU,O	100	119 of 119
GJB2	Congenital Deafness With Keratopachydermia And Constrictions Offingers And Toes, Ichthyosis, Hystrix-like, With Deafness , Keratitis-Ichthyosis-Deafness Syndrome, Keratoderma Hereditarium Mutilans,	AD,AR,X,XR,MU,D,G	99.89	413 of 419

	Kid Syndrome, Knuckle Pads-Leukonychia-Sensorineural Deafness-Palmoplantar Hyperkeratosis Syndrome			
GJB3	Erythrokeratodermia Variabilis	AD,AR,MU,D	100	39 of 39
GJB6	Hidrotic Ectodermal Dysplasia, Kid Syndrome	AD,AR,X,XR,MU,D,G	99.89	28 of 28
GTF2E2	Nonphotosensitive Trichothiodystrophy	AR	99.98	2 of 2
GTF2H5	Photosensitive Trichothiodystrophy	AR	100	8 of 8
HESX1	Septooptic Dysplasia, Combined Pituitary Hormone Deficiencies, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome	AD,AR	100	26 of 26
HRAS	Costello Syndrome, Epidermal Nevus, Somatic Giant Pigmented Hairy Nevus, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	34 of 34
HS6ST1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD	99.97	8 of 8
IL17RD	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD,AR	99.95	17 of 17
IL2RB	Immunodeficiency With Lymphoproliferation And Autoimmunity	AR	94.56	6 of 6
ITGB6	Amelogenesis Imperfecta Type Ih, Alopecia-Intellectual Disability Syndrome	AR	99.91	8 of 8
KDSR	Erythrokeratodermia Variabilis Et Progressiva	AR	99.87	9 of 10
KISS1R	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD,AR	99.41	42 of 43
KRAS	Aplasia Cutis Congenita With Epibulbar Dermoids, Cardiofaciocutaneous Syndrome, Ras-Associated Autoimmune Lymphoproliferative Syndrome Type IV, Schimmelpenning-Feuerstein-Mims Syndrome, Linear Nevus Sebaceus Syndrome, Noonan Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
KRT1	Bullous Erythroderma Ichthyosiformis Congenita Of Brocq, Ichthyosis Hystrix Curth-Macklin Type , Cyclic Ichthyosis With Epidermolytic Hyperkeratosis, Keratosis Palmoplantaris Striata III, Palmoplantar Keratoderma	AD,AR	100	67 of 67
KRT10	Bullous Erythroderma Ichthyosiformis Congenita Of Brocq, Erythroderma, Ichthyosiform, Ichthyosiform Congenital Reticular Erythroderma, Cyclic Ichthyosis With Epidermolytic Hyperkeratosis, Autosomal Dominant Epidermolytic Ichthyosis	AD,AR	98.32	69 of 76
KRT14	Dermatopathia Pigmentosa Reticularis, Epidermolysis Bullosa Herpetiformis, Epidermolysis Bullosa Simplex, Naegeli-Franceschetti-Jadassohn Syndrome	AD,AR	100	132 of 132
KRT16	Pachyonychia Congenita, Nonepidermolytic Palmoplantar Keratoderma, Epidermolytic Palmoplantar Keratoderma	AD	100	35 of 35
KRT2	Bullous Type Ichthyosis, Superficial Epidermolytic Ichthyosis	AD	100	18 of 18
KRT9	Epidermolytic Palmoplantar Keratoderma	AD	100	32 of 32
LIPN	Lamellar Ichthyosis	AR	99.62	1 of 1
LMNA	Autosomal Recessive Emery-Dreifuss Muscular Dystrophy, Autosomal Dominant Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Familial Partial Lipodystrophy, Restrictive Dermopathy	AD,AR	100	619 of 620
LOR	Vohwinkel Syndrome with Ichthyosis		87.37	10 of 10

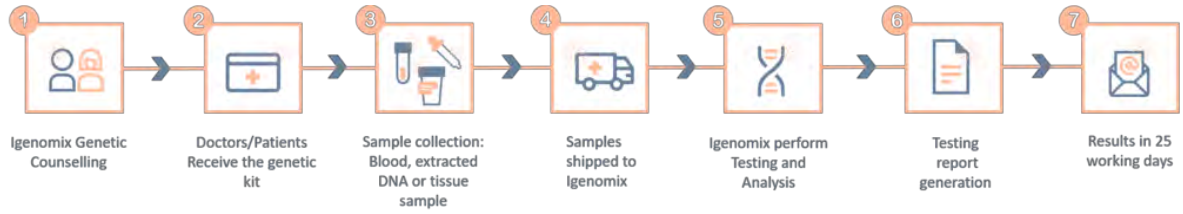
LORICRIN	Vohwinkel Syndrome, Keratoderma Hereditarium Mutilans With Ichthyosis, Progressive Symmetric Erythrokeratodermia	AD	na	na
LSS	Hypotrichosis, Alopecia-Intellectual Disability Syndrome, Hypotrichosis Simplex	AR	100	22 of 22
MAP2K1	Cardiofaciocutaneous Syndrome, Isolated Melorheostosis	AD	100	31 of 31
MAP2K2	Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100	37 of 37
MBTPS2	Ichthyosis Follicularis, Atrichia, And Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Palmoplantar Keratoderma, Mutilating, With Periorificial Keratoticplaques, Bresek Syndrome, Ichthyosis Follicularis-Alopecia-Photophobia Syndrome, Mutilating Palmoplantar Keratoderma With Periorificial Keratotic Plaques	X,XR,G	100	NA of NA
MPLKIP	Trichothiodystrophy	AR	100	13 of 13
MSMO1	Microcephaly, Congenital Cataract, And Psoriasiform Dermatitis	AR	99.78	4 of 4
NDNF	Hypogonadotropic Hypogonadism With Anosmia, Kallmann Syndrome	AD	99.33	NA of NA
NEK9	Arthrogryposis, Perthes Disease, And Upward Gaze Palsy, Lethal Congenital Contracture Syndrome, Nevus Comedonicus	AR	99.98	4 of 4
NIPAL4	Congenital Ichthyosis, Congenital Non-Bullous Ichthyosiform Erythroderma, Lamellar Ichthyosis	AR	88.73	34 of 37
NLRP3	Cinca Syndrome, Familial Cold Inflammatory Syndrome, Keratoendotheliitis Fugax Hereditaria, Muckle-Wells Syndrome	AD	100	152 of 152
NOD2	Inflammatory Bowel Disease, Synovitis, Granulomatous, With Uveitis And Cranial Neuropathies, Yao Syndrome, Blau Syndrome	AD,MU	100	97 of 97
NRAS	Epidermal Nevus, Somatic, Giant Pigmented Hairy Nevus, Neurocutaneous Melanosis, Ras-Associated Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	15 of 15
NSDHL	Congenital Hemidysplasia With Ichthyosiform Erythroderma And Limb, Ck Syndrome	X,XR,XD,G	100	NA of NA
NSMF	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome , Normosmic Congenital Hypogonadotropic Hypogonadism	AD	99.69	11 of 11
ORAI1	Immunodeficiency, Myopathy, Stormorken-Sjaastad-Langslet Syndrome, Tubular Aggregate Myopathy	AD,AR	91.93	20 of 22
OSMR	Primary Cutaneous Amyloidosis, Primary Cutaneous	AD	100	14 of 14
PEX1	Hearing Loss, Sensorineural, With Enamel Hypoplasia And Nail Defects , Peroxisome Biogenesis Disorder, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX10	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.76	29 of 32
PEX11B	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	90.29	7 of 7
PEX12	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	38 of 38
PEX13	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.98	11 of 12

PEX14	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	4 of 4
PEX16	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	17 of 17
PEX19	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	5 of 5
PEX2	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	99.89	17 of 17
PEX26	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	29 of 29
PEX3	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	9 of 9
PEX5	Adrenoleukodystrophy, Autosomal Neonatal Form, Cerebrohepatorenal Syndrome, Rhizomelic Chondrodysplasia Punctata, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	12 of 12
PEX6	Heimler Syndrome, Peroxisome Biogenesis Disorder (Zellweger), Autosomal Recessive Spinocerebellar Ataxia-Blindness-Deafness Syndrome, Infantile Refsum Disease , Neonatal Adrenoleukodystrophy , Zellweger Syndrome	AD,AR	99.94	105 of 108
PEX7	Peroxisome Biogenesis Disorder, Refsum Disease, Rhizomelic Chondrodysplasia Punctata	AR	99.21	47 of 53
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency	AR	100	26 of 26
PHYH	Refsum Disease	AR	100	34 of 34
PIGA	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Paroxysmal Nocturnal Hemoglobinuria	X,XR,G	97.98	NA of NA
PIGL	Zunich Neuroectodermal Syndrome, Chime Syndrome, Hyperphosphatasia-Intellectual Disability Syndrome	AR	86	11 of 13
PNPLA1	Congenital Ichthyosis, Congenital Non-Bullous Ichthyosiform Erythroderma	AR	99.9	57 of 57
PNPLA2	Neutral Lipid Storage Disease With Myopathy	AR	100	53 of 53
POMP	Keratosis Linearis With Ichthyosis Congenita And Sclerosing Keratoderma, Proteasome-Associated Autoinflammatory Syndrome, Keratosis Linearis-Ichthyosis Congenita-Sclerosing Keratoderma Syndrome	AD,AR	99.99	4 of 4
PROK2	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD	100	20 of 20
PROKR2	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD	100	64 of 64
PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
RIN2	Macrocephaly, Alopecia, Cutis Laxa, And Scoliosis, Rin2 Syndrome	AR	99.6	4 of 4
RNF113A	Nonphotosensitive Trichothiodystrophy	X,XD,G	99.7	NA of NA
SBDS	Aplastic Anemia, Shwachman-Diamond Syndrome	AR	100	77 of 79
SDR9C7	Autosomal Recessive Congenital Ichthyosis, Lamellar Ichthyosis	AR	99.99	10 of 10

SEMA3A	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD	100	29 of 29
SGPL1	Nephrotic Syndrome Type 14	AR	98.96	18 of 18
SHOC2	Noonan Syndrome-like Disorder With Loose Anagen Hair	AD	99.98	8 of 8
SLC27A4	Ichthyosis-Prematurity Syndrome	-	100	25 of 25
SLC29A3	Histiocytosis-Lymphadenopathy Plus Syndrome, Dysosteosclerosis, H Syndrome	AR	100	32 of 32
SLURP1	Mal De Meleda, Hereditary Palmoplantar Keratoderma, Gamborg-Mielsen Type	AR	100	24 of 24
SNAP29	Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratodermasyndrome, Cednik Syndrome	AR	100	13 of 13
SOX10	Peripheral Demyelinating Neuropathy, Central Dysmyelination, Waardenburg Syndrome	AD	99.74	139 of 147
SPINK5	Netherton Syndrome	AR	99.98	84 of 84
SPRY4	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.72	13 of 13
SRD5A3	Congenital Disorder Of Glycosylation Type Iq, Kahrizi Syndrome	AR	100	15 of 15
SRP54	Severe Congenital Neutropenia, Shwachman-Diamond Syndrome	AD,AR	99.95	8 of 8
ST14	Autosomal Recessive Congenital Ichthyosis	AR	100	10 of 10
STIM1	Stormorken Syndrome, Stormorken-Sjaastad-Langslet Syndrome, Tubular Aggregate Myopathy	AD,AR	100	28 of 28
STS	X-linked Ichthyosis	X,XR,G	100	NA of NA
SULT2B1	Autosomal Recessive Congenital Ichthyosis, Lamellar Ichthyosis	AR	100	6 of 6
SUMF1	Multiple Sulfatase Deficiency	AR	100	52 of 52
SYNE1	Arthrogryposis Multiplex Congenita, Myogenic Type, Emery-Dreifuss Muscular Dystrophy, Spinocerebellar Ataxia	AD,AR	99.99	193 of 193
SYNE2	Autosomal Dominant Emery-Dreifuss Muscular Dystrophy	AD	99.94	12 of 12
TACR3	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AR	99.97	40 of 40
TARS1	Nonphotosensitive Trichothiodystrophy	AR	99.94	NA of NA
TGM1	Lamellar Ichthyosis, Acral Self-Healing Collodion Baby, Bathing Suit Ichthyosis, Congenital Non-bullous Ichthyosiform Erythroderma	AR	100	206 of 208
TGM5	Acral Peeling Skin Syndrome	AR	100	26 of 26
TMEM43	Autosomal Dominant Emery-Dreifuss Muscular Dystrophy	AD	99.98	26 of 26
TMPRSS6	Iron-Refractory Iron Deficiency Anemia	AR	99.84	103 of 104
VIPAS39	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	15 of 15
VPS33B	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	62 of 62
WDR11	Hypogonadotropic Hypogonadism With Or Without Anosmia, Pituitary Stalk Interruption Syndrome	AD,AR	100	19 of 19

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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References

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