Autism and Attention Deficit Hyperactivity Disorder

Precision Panel

Overview

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction and restricted, repetitive patterns of behavior, interests and activities. These symptoms appear from early childhood and limit or impair everyday functioning. ASD may be an isolated and idiopathic condition or associated to genetic diseases such as Rett syndrome, neurofibromatosis, tuberous sclerosis and fragile X syndrome, among others. This increases the heritability of ASD to more than 90%. Attention deficit hyperactivity disorder (ADHD) is one of the most common neuropsychiatric disorders of childhood and adolescence, often persisting into adulthood. ADHD is characterized by symptoms of inattention, impulsiveness, restlessness, executive dysfunction and emotional dysregulation which lead to markedly decreased functioning. Often, ADHD shares comorbidity with other psychiatric conditions such as obsessive-compulsive disorder. ADHD is highly heritable and multifactorial; multiple genes and non-inherited factors contribute to the disorder. The risk of ADHD in parents and siblings of children with ADHD is increased 2-8 times with heritability of approximately 76%.

The Igenomix Autism and Attention Deficit Hyperactivity Disorder Precision Panel can serve as an accurate and directed diagnostic tool 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 Autism and Attention Deficit Hyperactivity Disorder is indicated in patients with a clinical suspicion or diagnosis of with or without the following manifestations:

- Persistent deficits in social communication and social interaction in multiple settings
- Restricted, repetitive patterns of behavior, interests, or activities
- Impairment of function
- Symptoms present in early developmental period
- Symptoms not explained by intellectual disability
- Language delays and deviation
- Hyperactivity and distractibility
- Difficulty performing daily tasks, lack of concentration
- Forgetful in daily activities
- Excessive talking

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient. Improve diagnostic criteria, natural history studies and novel therapeutic options.
- Early initiation of treatment with a multidisciplinary team in the form of behavioral, educational and psychological therapies, which have proven to be the most effective for ASD.
- In the case of ADHD, environmental restructuring and behavioral therapy as well as developments in behavioral parent training (BPT) and behavioral classroom management (BCM). Medical care with stimulants is also considered as a first-line treatment.
- 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**
AARS1	Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AD,AR	99,07	30 of 30
ABCA7	Alzheimer Disease	AD	99,99	159 of 159
ABCD1	Adrenoleukodystrophy	X,XR,G	100	-
ACHE	Colonic Pseudo-Obstruction, Myasthenic Syndrome	-	98,34	4 of 4
ACSL4	Intellectual Disability, Alport Syndrome, Midface Hypoplasia, Elliptocytosis	X,XD,G	99,97	-
ACTL6A	Intellectual Disability	-	99,98	3 of 3
ACTL6B	Epileptic Encephalopathy, Intellectual Developmental Disorder, Speech And Ambulation Defects	AD,AR	100	21 of 21
ADA	Immunodeficiency, Omenn Syndrome	AR	100	97 of 98
ADCY3	Joubert Syndrome, Hypothyroidism	AR	97,98	7 of 7
ADNP	Helsmoortel-Van Der Aa Syndrome, Adnp Syndrome	AD	99,91	90 of 90
ADSL	Adenylosuccinate Lyase Deficiency	AR	100	59 of 59
AFF2	Intellectual Disability	X,XR,G	99,5	-
AGAP2	Fragile X Syndrome, Spinal Canal Intradural Extramedullary Neoplasm	-	95,15	7 of 7
AGO1	Corpus Callosum Agenesis, Squamous Cell Carcinoma, Cartilage-Hair Hypoplasia, Retinitis Pigmentosa, Autism Spectrum Disorder	-	100	7 of 7
AGO4	Cartilage-Hair Hypoplasia	-	98,73	4 of 4
AGTR2	Intellectual Disability	-	99,94	-
AHDC1	Xia-Gibbs Syndrome, Intellectual Disability, Obstructive Sleep Apnea	AD	99,87	41 of 43
AKAP9	Long Qt Syndrome, Brugada Syndrome, Romano-Ward Syndrome	AD	98,34	43 of 46
AKT1	Breast Cancer, Colorectal Cancer, Cowden Syndrome, Proteus Syndrome, Meningioma	AD	100	6 of 6
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95,41	65 of 69
ALG13	Epileptic Encephalopathy, Congenital Disorder Of Glycosilation, Intellectual Disability	X,XR,XD,G	99,62	-
ALKBH8	Intellectual Developmental Disorder	AR	99,2	2 of 2
AMPD1	Myopathy, Adenosine Monophosphate Deaminase Deficiency	AR	100	10 of 10
AMT	Glycine Encephalopathy	AR	99,98	94 of 96
ANK2	Cardiac Arrhythmia, Romano-Ward Syndrome	AD	99,98	130 of 130

ANK3	Intellectual Disability, Sleep Disturbance	AR	99,76	22 of 23
ANKRD11	Kbg Syndrome, 16q24.3 Microdeletion Syndrome	AD	99,6	119 of 124
ANXA1	Hairy Cell Leukemia, Brain Edema, Rheumatoid Arthritis, Colon Adenocarcinoma, Squamous Cell Carcinoma	-	99,99	1 of 2
AP1S2	Intellectual Disability, Fried Syndrome, Dandy-Walker Malformation, Basal Ganglia Disease, Seizures	X,XR,G	84,15	-
AP2S1	Hypocalciuric Hypercalcemia	AD	90	6 of 6
AP3B2	Epileptic Encephalopathy	AR	99,95	11 of 12
APBB1	Niemann-Pick Disease, Alzheimer Disease	-	92,31	1 of 1
APC2	Cortical Dysplasia, Sotos Syndrome	AR	94,97	11 of 11
APH1A	Immunodeficiency, Alzheimer Disease, Cerebral Amyloid Angiopahty, Parkinson Disease	-	100	3 of 3
ARF1	Periventricular Nodular Heterotopia	AD	100	3 of 3
ARHGEF6	Intellectual Disability	-	97,04	-
ARHGEF9	Hyperekplexia, Epilepsy	X,XR,G	100	-
ARID1B	Coffin-Siris Syndrome, 6q25 Microdeletion Syndrome	AD	93,87	226 of 238
ARID2	Coffin-Siris Syndrome	AD	99,97	17 of 17
ARNT2	Webb-Dattani Syndrome, Septo-Optic Dysplasia Spectrum	AR	100	5 of 5
ARV1	Epileptic Encephalopathy	AR	100	3 of 3
ARVCF	22q11.2 Deletion Syndrome	-	99,95	2 of 2
ARX	Corpus Callosum Agenesis, Epileptic Encephalopathy, Lissencephaly, Intellectual Disability, Partington Syndrome, West Syndrome	X,XR,G	81,92	-
ASAP2	Bulbar Polio, Epiphyseal Dysplasia	-	99,83	4 of 4
ASH1L	Intellectual Disability	AD	98,78	57 of 57
ASPM	Microcephaly	AR	99,74	221 of 222
ASTN2	Bardet-Biedl Syndrome, Limb Girdle Muscular Dystrophy, Myopathy	-	95,58	6 of 12
ASXL3	Bainbridge-Ropers Syndrome, Severe Feeding Difficulties, Microcephaly	AD	95,96	77 of 81
ATP10A	Epilepsy, Ventricular Septal Defect, Esophageal Atresia, Chromosome 15q11-Q13 Duplication Syndrome	-	98,27	5 of 5
ATP2B2	Deafness	AR	100	12 of 12
ATP6V1A	Cutis Laxa, Epileptic Encephalopathy	AD,AR	99,98	9 of 9
ATRX	Alpha-Thalassemia Myelodysplasia Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98,5	-
AUTS2	Intellectual Disability, Autism Spectrum Disorder	AD	99,63	9 of 17
AVPR1A	Amusia, Hepatorenal Syndrome, Diabetes Insipidus, Borderline Personality Syndrome, Macronodular Adrenal Hyperplasia	-	99,84	1 of 1
BAZ1B	Williams Syndrome	-	99,05	5 of 5
BAZ2B	Corpus Callosum Agenesis	-	99,29	9 of 9
BCKDK	Branched-Chain Ketoacid Dehydrogenase Kinase Deficiency	-	99,91	6 of 6
BCL11A	Intellectual Developmental Disorder	AD	99,9	22 of 22
BCORL1	Shukla-Vernon Syndrome, Intellectual Disability	X,XR,G	98,77	-
BCR	Leukemia, 22q11.2 Microdeletion Syndrome	MU,P	97,78	-
BDNF	Ondine Syndrome, Wagr Syndrome	-	99,96	7 of 7
BMPR1A	Polyposis Syndrome	AD	100	124 of 127
BPTF	Neurodevelopmental Disorder, Dysmorphic Facies, Distal Limb Anomalies, 17q24.2 Microdeletion Syndrome, Intellectual Disability	AD	94,31	12 of 15
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Noonan Syndrome, Craniopharyngioma	AD	100	80 of 80

BRSK2	Paraneoplastic Limbic Encephalitis, Myasthenic Syndrome, Intelectual Disability	-	98,91	10 of 10
BTAF1	Leukemia, Epilepsy, Kbg Syndrome, Epiphyseal Chondrodysplasia	-	99,8	1 of 1
C12ORF4	Intellectual Disability	AR	-	-
C12ORF57	Craniofacial Dysmorphism, Ocular Coloboma, Corpus Callosum Agenesis, Aortic Dilatation, Temtamy Syndrome	AR	-	-
CACNA1A	Epileptic Encephalopathy, Spinocerebellar And Paroxisomal Ataxia, Torticollis Of Infancy, Hemiplegic Migraine	AD	96,13	249 of 266
CACNA1B	Neurodevelopmental Disorder, Seizures, Hyperkinetic Movements, Epileptic Encephalopathy	AR	95,83	7 of 7
CACNA1C	Brugada Syndrome, Timothy Syndrome, Romano-Ward Syndrome	AD	99,8	85 of 85
CACNA1D	Seizures, Sinoatrial Node Dysfunction, Deafness, Primary Hyperaldosteronism, Neurological Abnormalities	AD,AR	100	18 of 18
CACNA1E	Epileptic Encephalopathy	AD	99,94	25 of 25
CACNA1H	Hyperaldosteronism, Epilepsy	AD	98,05	71 of 71
CACNA2D3	Alkuraya-Kucinskas Syndrome, Zimmermann-Laband Syndrome	-	95,98	6 of 6
CACNB2	Brugada Syndrome	AD	99,84	32 of 34
CAPRIN1	Moyamoya Angiopathy, Amyotrophic Lateral Sclerosis	-	99,97	2 of 2
CARS1	Microcephaly, Developmental Delay, Brittle Hair Syndrome	AR	100	7 of 7
CASK	Anemia, Fg Syndrome, Intellectual Disability, Microcephaly, Epileptic Encephalopathy	X,XR,XD,G	99,98	-
CASZ1	Dystonia, Chromosome 1p36 Deletion Syndrome, Neuroblastoma, Speech Disorder, Dilated Cardiomyopathy	-	90,5	6 of 6
CC2D1A	Intellectual Disability	AR	100	7 of 7
CCNG1	Multiple Cancer Types, Beckwith-Wiedemann Syndrome	-	99,95	-
CCT4	Sensory And Peripheral Neuropathy, Peroxisome Biogenesis Disorder, Dystonia	-	99,95	2 of 2
CDC42BPB	Myotonic Dystrophy, Epidermolysis Bullosa Simplex, Autism Spectrum Disorder	-	100	5 of 5
CDH13	VACTERL Association, Adenomatous Polyposis, Seminoma, Lung Cancer	-	99,94	5 of 5
CDH2	Corpus Callosum Agenesis, Arrhythmogenic Right Ventricular Dysplasia	AD	99,98	16 of 16
CDK19	Epileptic Encephalopathy	AD	99,81	1 of 1
CDK8	Intellectual Developmental Disorder, Hypotonia	AD	99,89	8 of 8
CDKL5	Epileptic Encephalopathy, Rett Syndrome, West Syndrome	X,XD,G	99,92	-
CDON	Holoprosencephaly, Pituitary Stalk Interruption Syndrome	AD	100	15 of 15
CELF4	Nephrotic Syndrome, Frontotemporal Dementia, Epilepsy, Autism	-	99,84	-
CEP135	Microcephaly	AR	99,48	7 of 8
CEP41	Joubert Syndrome	AR	100	17 of 17
CERT1	Intellectual Disability	AD	99,98	8 of 8
CGNL1	Aromatase Excess Syndrome, Estrogen Excess	-	100	3 of 3
CHAMP1	Intellectual Disability	AD	99,85	17 of 17
CHD1	Pilarowski-Bjornsson Syndrome, Intellectual Disability, Autism, Speech Apraxia, Craniofacial Dysmorphism	AD	99,06	8 of 8
CHD2	Epileptic Encephalopathy, Lennox-Gastaut Syndrome, Myoclonic-Astastic Epilepsy	AD	98,91	103 of 103
CHD3	Snijders Blok-Campeau Syndrome	AD	97,93	30 of 30
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism, Kallmann Syndrome, Omenn Syndrome	AD	96,25	823 of 896
CHD8	Autism	AD	99,91	119 of 120
CHMP1A	Pontocerebellar Hypoplasia	AR	100	4 of 4
CHRNA7	Chromosome 15q13.3 Microdeletion Syndrome	AD	82,09	2 of 2
CIB2	Deafness, Usher Syndrome	AR	99,95	16 of 17
CIC	Intellectual Disability	AD	63,02	11 of 13

CLASP1	Lowry-Wood Syndrome, Roifman Syndrome, Theileriasis	-	99,98	4 of 4
CLCN4	Intellectual Disability	X,XR,XD,G	99,69	-
CLIP2	Williams Syndrome	-	99,99	1 of 1
CLTC	Intellectual Disability, Epileptic Encephalopathy	AD	98,81	14 of 14
CNKSR2	Intellectual Disability, Epileptic Encephalopathy	X,G	99,11	-
CNOT3	Intellectual Developmental Disorder, Speech Delay, Autism, Dysmorphic Facies	AD	100	25 of 25
CNR1	Anxiety, Substance Dependence	-	99,98	17 of 17
CNTN4	Chromosome 3pter-P25 Deletion Syndrome, Coffin-Siris Syndrome, Spinocerebellar Ataxia, Chromosome 14q11-Q22 Deletion Syndrome, Autism Spectrum Disorder	-	100	5 of 6
CNTN5	Chromosome 3pter-P25 Deletion Syndrome, Coffin-Siris Syndrome, Autism Spectrum Disorder, Cyclothymic Disorder, Atrial Septal Defect	-	99,69	3 of 3
CNTN6	Autism, Chromosome 3pter-P25 Deletion Syndrome,	-	99,95	9 of 9
CNTNAP2	Pitt-Hopkins-Like Syndrome	AR	99,91	39 of 41
CNTNAP3	Intellectual Disability, Cloacal Exstrophy, Cyclothymic Disorder, Ectodermal Dysplasia, Cleft Lip/Palate	-	84,58	0 of 1
CNTNAP4	Kagami-Ogata Syndrome	-	99,97	2 of 2
CNTNAP5	Posterior Cortical Atrophy, Dyslexia, Austim	-	99,98	4 of 4
COMT	Panic Disorder, Schizophrenia, 22q11.2 Deletion Syndrome	AD	99,98	5 of 5
CORO1A	Immunodeficiency	AR	93	9 of 9
CPLX1	Epileptic Encephalopathy, Wolf-Hirschhorn Syndrome, Myoclonic Epilepsy	AD,AR	99,81	3 of 3
CRBN	Intellectual Disability	AR	100	4 of 4
CREBBP	Menke-Hennekam Syndrome, Rubinstein-Taybi Syndrome	AD	100	318 of 318
CRKL	22q11.2 Microdeletion Syndrome	-	99,93	5 of 6
CSDE1	Poliomyelitis, Bulbar Polio, Lymphadenopathy, Diamond-Blackfan Anemia	-	99,98	18 of 18
CSGALNACT1	Skeletal Dysplasia, Joint Laxity	AR	100	4 of 5
CSMD1	Autism Spectrum Disorder, Schizophrenia, Substance Dependance	-	99,98	21 of 27
CSNK2A1	Okur-Chung Neurodevelopmental Syndrome	AD	99,95	23 of 23
CTCF	Intellectual Disability, Developmental Delay, Microcephaly	AD	96,6	39 of 41
CTNNB1	Exudative Vitreoretinopathy, Hepatocellular Carcinoma, Medulloblastoma, Intellectual Disability, Pilomatrixoma, Craniopharyngioma, Desmoid Tumor, Pediatric Hepatocellular Carcinoma	AD,AR	100	63 of 63
CTNND2	Myoclonic Epilepsy, Monosomy 5p	-	94,3	10 of 12
CTTNBP2	Noonan Syndrome, Microphthalmia, Actinobacillosis	-	99,03	34 of 34
CUL3	Pseudohypoaldosteronism	AD	99,88	25 of 34
CUL7	3m Syndrome	AR	99,94	92 of 92
CUX1	Global Developmental Delay, Intellectual Development	AD	97,72	5 of 6
CXORF56	Intellectual Disability	X,G	-	-
CYFIP1	Fragile X Syndrome, Cataract, Dyscalculia, Prader-Willi Syndrome, Autism Spectrum Disorder	-	99,94	1 of 1
CYFIP2	Epileptic Encephalopathy	AD	100	8 of 8
DALRD3	Epileptic Encephalopathy	AR	97,17	-
DDX3X	Intellectual Developmental Disorder, Hypotonia	X,XR,XD,G	99,03	-
DDX6	Intellectual Developmental Disorder, Impaired Language, Dysmorphic Facies	-	100	5 of 5
DEAF1	Dyskinesia, Seizures, Intellectual Developmental Disorder, Intellectual Disability, Epilepsy, Smith-Magenis Syndrome	AD,AR	93,55	42 of 42
DENR	Optic Atrophy, Cranial Nerve Disease, 3-Methylglutaconic Aciduria, Parkinson Disease	-	99,63	1 of 2
DGAT2L6	Epilepsy, Centrotemporal Spikes, Cornelia De Lange Syndrome	-	99,92	-

DHCR7	Smith-Lemli-Opitz Syndrome	AR	100	217 of 217
DHDDS	Developmental Delay, Seizures, Retinitis Pigmentosa, Epileptic Encephalopathy	AD,AR	96,32	8 of 8
DHTKD1	2-Aminoadipic 2-Oxoadipic Aciduria, Charcot-Marie-Tooth Disease	AD,AR	99,94	25 of 25
DIP2A	Dyslexia, Reading Disorder, Autism Spectrum Disorder, Long Qt Syndrome	-	100	5 of 5
DIP2C	Spastic Hemiplegia, Fraser Syndrome, Spinocerebellar Ataxia, Hemochromatosis	-	99,95	4 of 4
DISC1	Microcephaly, Polymicrogyria, Corpus Callosum Agenesis	-	97,88	16 of 17
DISP1	Holoprosencephaly	-	100	10 of 10
DLG2	Schizophrenia, Renal Oncocytoma, Tarp Syndrome, Autism Spectrum Disease	-	100	7 of 8
DLG3	Intellectual Disability	X,XR,G	100	-
DLG4	Intellectual Developmental Disorder	AD	99,83	13 of 13
DLGAP1	Transient Tic Disorder, Obsessive-Compulsive Disorder, Trichotillomania, Febrile Seizures, Myasthenic Syndrome	-	99,76	8 of 8
DLGAP2	Lung Cancer, Obsessive-Compulsive Disorder, Childhood Disintegrative Disease, Anxiety Disorder, Ceroid Lipofuscinosis	-	99,97	14 of 14
DLL1	Neurodevelopmental Disorder, Holoprosencephaly	AD	99,83	15 of 15
DLX3	Amelogenesis Imperfecta, Trichodentoosseous Syndrome	AD	100	10 of 10
DMD	Cardiomyopathy, Becker And Duchenne Muscular Dystrophy, Intellectual Disability	X,XR,G	99,96	-
DMPK	Steinert Myotonic Dystrophy	AD	99,83	3 of 3
DNAJC12	Hyperphenylalaninemia	AR	81,82	10 of 10
DNM1	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	94,8	30 of 30
DNMT3A	Heyn-Sproul-Jackson Syndrome, Tatton-Brown-Rahman Syndrome, Pheochromocytoma, Paraganglioma, Intellectual Disability, Facial Dysmorphism	AD	99,95	67 of 68
DOCK3	Neurodevelopmental Disorder, Hypotonia, Ataxia	AR	99,94	7 of 8
DOCK4	Deafness, Dyslexia, Placenta Accreta, Usher Syndrome	-	99,94	3 of 3
DOCK8	Hyperimmunoglobulin E-Recurrent Infection Syndrome, Immunodeficiency	AR	99,92	106 of 114
DPH1	Developmental Delay, Short Stature, Dysmorphic Features, Craniofacial Dysplasia, Ectodermal Anomalies, Intellectual Disability	AR	100	8 of 8
DPP10	Asthma, Schizophrenia, Bipolar Disorder, Autism Spectrum Disorder	-	98,97	-
DPP6	Intellectual Disability, Ventricular Fibrillation, Microcephaly	AD	97,03	23 of 28
DPYD	Dihydropyrimidine Dehydrogenase Deficiency, 1p21.3 Microdeletion Syndrome	AR	100	74 of 75
DPYSL2	Ceroid Lipofuscinosis, Alzheimer Disease, Biopolar Disorder, Schizophrenia, Austism Spectrum Disorder	-	96,1	4 of 4
DRD4	Attention Deficit-Hyperactivity Disorder	AD	83,4	3 of 3
DRD5	Attention Deficit-Hyperactivity Disorder, Blepharospasm	AD	98,8	-
DSCAM	Intellectual Disability, Down Syndrome, Hirschsprung Disease, Enterokinase Deficiency, Heart Disease	-	99,87	40 of 40
DYM	Dyggve-Melchior-Clausen Disease, Smith-Mccort Dysplasia	AR	90	37 of 37
DYNC1H1	Charcot-Marie-Tooth Disease, Intellectual Disability, Spinal Muscular Atrophy	AD	100	104 of 104
DYNC1I2	Neurodevelopmental Disorder, Microcephaly, Structural Brain Anomalies	AR	99,97	3 of 3
DYRK1A	Intellectual Disability	AD	99,85	78 of 81
EBF3	Hypotonia, Ataxia, Delayed Development	AD	100	25 of 25
EEF1A2	Epileptic Encephalopathy, Intellectual Disability	AD	100	14 of 14
EFR3A	Autism Spectrum Disorder, Ciliary Diskinesia	-	99,97	1 of 1
EHMT1	Kleefstra Syndrome	AD	98,58	58 of 75
EIF3G	Narcolepsy	-	94,09	2 of 2
ELAVL3	Paraneoplastic Limbic Encephalitis, Sigmoid Disease, Peripheral Neuropathy, Lambert-Eaton Myasthenic Syndrome	-	99,99	2 of 2

ELN	Cutis Laxa, Supravalvular Aortic Stenosis, Williams-Beuren Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection	AD	99,99	95 of 96
ELP4	Aniridia	AD	96,31	1 of 4
EN2	Autism Spectrum Disorder, Hyperinsulinemic Hypoglycemia, Cerebellar Hypoplasia, Charcot Marie Tooth Disease	-	80,73	1 of 2
EP300	Menke-Hennekam Syndrome, Rubinstein-Taybi Syndrome	AD	100	109 of 109
EP400	Ossifying Fibromyxoid Tumor, Epilepsy	-	99,91	7 of 7
EPCAM	Diarrhea, Lynch Syndrome	AR	99,94	52 of 70
ETFB	Acyl-Coa Dehydrogenase Deficiency	AR	100	21 of 21
EXT2	Seizures, Scoliosis, Macrocephaly Syndrome, Potocki-Shaffer Syndrome	AD,AR	100	251 of 254
FAN1	Interstitial Nephritis, Lynch Syndrome	AR	99,97	22 of 22
FBN1	Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Microspherophakia, Short Stature, Shprintzen-Goldberg Syndrome	AD	100	2836 of 2845
FBXO11	Intellectual Developmental Disorder, Dysmorphic Facies, Behavioral Abnormalities	AD	95,81	42 of 45
FBXW11	Neurodevelopmental-Jaw-Eye-Digital Syndrome, Intellectual Disability	AD	99,89	10 of 10
FGD1	Aarskog-Scott Syndrome	X,XR,G	98,95	-
FGF12	Epileptic Encephalopathy	AD	99,98	4 of 6
FGF8	Hypogonadotropic Hypogonadism, Holoprosencephaly, Kallmann Syndrome	AD	98,36	38 of 38
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Kallmann Syndrome, Pfeiffer Syndrome, Trigonocephaly, Holoprosencephaly, Hypogonadotropic Hypogonadism	AD	100	279 of 280
FGFR3	Achondroplasia, Developmental Delay, Acanthosis Nigricans, Camptodactyly, Crouzon Syndrome, Hypochondroplasia, Lacrimoauriculodentodigital Syndrome, Thanatophoric Dysplasia, Muenke Syndrome, Saethre-Chotzen Syndrome	AD,AR	99,89	77 of 78
FLCN	Birt-Hogg-Dube Syndrome, Potocki-Lupski Syndrome	AD	100	200 of 205
FLI1	Bleeding Disorder, Jacobsen Syndrome, Paris-Trousseau Thrombocytopenia	AD,AR	100	7 of 7
FLII	Smith-Magenis Syndrome	-	99,98	3 of 3
FMR1	Fragile X Intellectual Disability Syndrome, Tremor/Ataxia Syndrome, Xq27.3q28 Duplication Syndrome	X,XD,G	99,8	-
FOLR1	Neurodegeneration	AR	100	19 of 23
FOXG1	Rett Syndrome, 14q12 Microdeletion Syndrome, Foxg1 Syndrome	AD	88,71	93 of 109
FOXH1	Holoprosencephaly	-	98,72	30 of 33
FOXP1	Intellectual Disability, Language Impairment, Autism, Speech Delay, Mild Dysmorphism Syndrome	AD	100	63 of 80
FOXP2	Speech-Language Disorder, Childhood Apraxia Of Speech	AD	100	17 of 17
FRMPD4	Intellectual Disability	X,XR,G	99,71	-
FTSJ1	Intellectual Disability	X,XR,G	100	-
GABRA1	Epileptic Encephalopathy, Dravet Syndrome	AD	100	45 of 46
GABRA2	Alcohol Dependence, Epileptic Encephalopathy	AD,MU	99,08	3 of 3
GABRA5	Epileptic Encephalopathy	AD	99,94	9 of 9
GABRB2	Epileptic Encephalopathy	AD	99,19	16 of 19
GABRB3	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	100	54 of 62
GABRD	Epilepsy, 1p36 Deletion Syndrome, Febrile Seizures	AD	95,23	3 of 3
GABRG2	Epileptic Encephalopathy, Dravet Syndrome, Rolandic Epilepsy	AD	99,67	53 of 53
GABRG3	Angleman Syndrome, Asperger Syndrome, Amphetamine Abuse, Albinism	-	100	2 of 2
GALC	Krabbe Disease	AR	99,38	252 of 254

GAMT	Cerebral Creatine Deficiency Syndrome, Guanidinoacetate Methyltransferase Deficiency	AR	99,92	60 of 60
GAS1	Holoprosencephaly	-	95,93	6 of 6
GATA4	Atrial Septal Defect, Testicular Anomalies, Tetralogy Of Fallot, 8p23.1 Microdeletion Syndrome	AD	94,69	108 of 130
GATM	Cerebral Creatine Deficiency Syndrome, Fanconi Renotubular Syndrome	AD,AR	99,98	21 of 21
GDI1	Intellectual Disability	X,XD,G	100	-
GFAP	Alexander Disease	AD	99,98	143 of 143
GGNBP2	Chromosome 17q12 Deletion Syndrome	-	99,65	4 of 4
GIGYF1	Autism Spectrum Disease, Tourette Syndrome	-	100	10 of 10
GIGYF2	Parkinson Disease	AD	99,88	49 of 49
GJA5	Atrial Fibrillation, Chromosome 1q21.1 Deletion Syndrome, Tetralogy Of Fallot	AD	99,88	13 of 13
GJA8	Cataract, Chromosome 1q21.1 Deletion Syndrome, Cataract-Microcornea Syndrome	AD	99,2	72 of 73
GLI2	Holoprosencephaly, Pallister-Hall Syndrome, Pituitary Hormone Deficiencies	AD	98,38	83 of 88
GLUD1	Hyperinsulinemic Hypoglycemia	AD	99,98	39 of 39
GNAI1	Cerebellar Astrocytoma, Corpus Callosum Agenesis	-	99,42	8 of 8
GNAQ	Sturge-Weber Syndrome	AD	99,97	3 of 3
GNB5	Intellectual Developmental Disorder, Language Delay, Attention Deficit, Hyperactivity Disorder, Cognitive Impairment	AR	100	13 of 13
GNE	Nonaka Myopathy, Sialuria	AD,AR	99,97	248 of 253
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Alloimmune Thrombocytopenia	AR	74,08	26 of 50
GPC4	Keipert Syndrome, Simpson-Golabi-Behmel Syndrome, Wilms Tumor	AD,X,XR,G	98,43	-
GPHN	Hyperekplexia, Molybdenum Cofactor Deficiency	AD,AR	99,2	6 of 6
GRIA1	Depression, Cortical Dysplasia, Tarp Syndrome, Limbic Encephaliis, Intelectual Disability	-	99,92	5 of 5
GRIA2	Neurodevelopmental Disorder, Language Impairment, Behavioral Abnormalities	AD	98,78	20 of 20
GRIA3	Intellectual Disability	X,XR,G	98,39	-
GRIA4	Neurodevelopmental Disorder, Seizures, Gait Abnormalities	AD	99,94	5 of 5
GRID1	Rett Syndrome, Pitt-Hopkins Syndrome, Schizophrenia	-	99,96	3 of 3
GRIK2	Intellectual Disability	AR	96,98	5 of 6
GRIK5	Brugada Syndrome, Epilepsy, Schizophrenia, Bipolar Disorder, Intellectual Disability	-	94,01	4 of 4
GRIN1	Neurodevelopmental Disorder, Hyperkinetic Movements, Seizures	AD,AR	100	43 of 43
GRIN2A	Speech Dyspraxia, Intellectual Disability, Epileptic Encephalopathy, Rolandic Epilepsy	AD	100	143 of 143
GRIN2B	Epileptic Encephalopathy, Intellectual Disability, West Syndrome	AD	99,99	108 of 108
GRIN2D	Epileptic Encephalopathy	AD	79,74	17 of 18
GRIP1	Fraser Syndrome	AR	100	17 of 17
GRPR	Agoraphobia, Autism	-	100	-
GSPT2	Autism, Seizure Disorder, Leukodystrophy, Intellectual Disability	-	99,94	-
GTF2I	Williams Syndrome	-	63,79	-
GTF2IRD1	Williams Syndrome	-	99,98	1 of 1
HCFC1	Methylmalonic Acidemia, Homocysteinemia, Intellectual Disability	X,XR,G	99,81	-
HCN1	Epileptic Encephalopathy, Febrile Seizures	AD	98,43	42 of 43
HDAC4	2q37 Microdeletion Syndrome	-	100	10 of 10
HDAC8	Cornelia De Lange Syndrome, Wilson-Turner Syndrome	X,XD,G	99,78	-
HDC	Gilles De La Tourette Syndrome	AD	100	4 of 4

HDLBP	Epilepsy, Chromosome 2q37 Deletion Syndrome	-	100	2 of 2
HECTD4	Bone Angioendothelial Sarcoma, Bone Epithelioid Hemangioma, Cataract	-	99,92	3 of 3
HECW2	Neurodevelopmental Disorder, Hypotonia, Seizures, Absent Language	AD	99,85	13 of 13
HERC2	Intellectual Disability, Prader-Willi Syndrome	AD,AR	98,91	9 of 9
HESX1	Hypothyroidism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD,AR	100	26 of 26
HIRA	22q11.2 Deletion Syndrome	-	99,99	5 of 5
HIVEP2	Intellectual Disability	AD	99,88	22 of 22
HIVEP3	Meckel Syndrome, Autism Spectrum Disorder	-	99,74	14 of 14
HMGN1	Down Syndrome, Cockayne Syndrome	-	97,32	-
HNF1B	Diabetes Mellitus, 17q12 Microdeletion Syndrome	AD	100	219 of 220
HNRNPH2	Intellectual Disability	X,XD,G	-	-
HNRNPU	Epileptic Encephalopathy, 1q44 Microdeletion Syndrome	AD	99,8	36 of 36
HOXA1	Athabaskan Brainstem Dysgenesis Syndrome	-	99,98	6 of 6
HOXA2	Microtia, Hearing Impairment, Cleft Palate	AD,AR	99,93	5 of 5
HPRT1	Gout, Lesch-Nyhan Syndrome, Hypoxanthine Guanine Phosphoribosyltransferase Partial Deficiency	X,XR,G	99,86	-
HRAS	Costello Syndrome, Epidermal Nevus, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	34 of 34
HSPG2	Dyssegmental Dysplasia, Schwartz-Jampel Syndrome, Schwartz-Jampel Syndrome	AR	99,41	68 of 69
HUWE1	Intellectual Disability	X,G	99,41	-
ICA1	Diabetes Mellitus, Insulinoma, Cataract	-	99,6	1 of 1
IFNG	Aplastic Anemia, Immunodeficiency	AD,AR	99,77	-
IGF1	Insulin-Like Growth Factor I Deficiency, Growth Delay	AR	100	7 of 8
IKBKG	Ectodermal Dysplasia, Immunodeficiency, Incontinentia Pigmenti	X,XR,XD,G	38,16	-
IL1RAPL1	Intellectual Disability	X,XR,G	99,78	-
ILF2	Endodermal Sinus Tumor	-	99,95	1 of 1
IMMP2L	Tic Disorder, Deafness, Tourette Syndrome, Apraxia Of Speech, Nephropathy	-	99,83	2 of 3
INTS6	Kbg Syndrome	-	99,33	4 of 4
IPW	Prader-Willi Syndrome	AD	-	-
IQSEC1	Intellectual Developmental Disorder, Short Stature, Behavioral Abnormalities	AR	99,92	3 of 3
IQSEC2	Intellectual Disability, Microduplication Xp11.22p11.23 Syndrome, Microcephaly, Smith-Magenis Syndrome	X,XR,XD,G	99,73	-
IRF2BPL	Neurodevelopmental Disorder, Abnormal Movements, Loss Of Speech, Seizures	AD	95,01	24 of 25
ITGB3	Glanzmann Thrombasthenia, Thrombocytopenia	AD,AR	99,44	178 of 179
JARID2	Tetralogy Of Fallot, Lymphedema, Weaver Syndrome	-	99,98	15 of 15
JMJD1C	22q11.2 Deletion Syndrome	-	99,09	27 of 27
JRK	Epilepsy	-	-	-
KANSL1	Koolen-De Vries Syndrome	AD	96,03	22 of 27
KAT2B	Holt-Oram Syndrome, Spinocerebellar Ataxia, Chromosome 16q13.3 Deletion Syndrome, Hemangioma Of Spleen, Microphthalmia	-	94,02	4 of 4
KAT6A	Arboleda-Tham Syndrome, Intellectual Disability, Craniofacial Anomalies	AD	99,89	66 of 68
KAT8	Li-Ghorgani-Weisz-Hubshman Syndrome	AD	99,97	1 of 1
KATNAL2	Microcystic Meningioma	-	96,12	5 of 5
KCNA2	Epileptic Encephalopathy	AD	99,86	23 of 23
KCNAB2	1p36 Deletion Syndrome	-	79	3 of 3

KCNB1	Epileptic Encephalopathy	AD	99,95	55 of 55
KCNJ10	Enlarged Vestibular Aqueduct, Pendred Syndrome, Seizures, Sensorineural Deafness, Ataxia, Intellectual Disability, East Syndrome	AR	93,53	27 of 32
KCNMA1	Cerebellar Atrophy, Developmental Delay, Seizures, Epilepsy, Paroxysmal Dyskinesia, Liang-Wang Syndrome	AD,AR	99,98	24 of 26
KCNQ2	Epileptic Encephalopathy	AD	99,94	333 of 334
KCNQ3	Epilepsy	AD	97,94	40 of 40
KCNS3	Epilepsy, Febrile Seizures	-	99,63	2 of 2
KCTD13	Osteoarthritis, Schizophrenia, Autism	-	82,35	-
KDM3B	Diets-Jongmans Syndrome	AD	96,72	18 of 20
KDM4C	Spermatogenic Failure, Brain Stem Cancer	-	99,94	1 of 1
KDM5B	Intellectual Disability	AR	97,44	41 of 41
KDM5C	Intellectual Disability	X,XR,G	100	-
KDM6A	Kabuki Syndrome	AD,X,XD,G	99,98	-
KDM6B	Neurodevelopmental Disorder	AD	99,98	20 of 20
KIAA1586	Schizophrenia, Potocki-Lupski Syndrome	-	95,56	-
KIF11	Lymphedema, Microcephaly, Chorioretinopathy	AD	99,78	82 of 89
KIF14	Meckel Syndrome, Microcephaly	AR	99,84	18 of 18
KIRREL3	Intellectual Disability, Jacobsen Syndrome, Nephrotic Syndrome	-	99,08	14 of 14
KLLN	Cowden Syndrome	-	97,52	9 of 9
KMT2A	Short Stature, Facial Dysmorphism, Developmental Delay, Cornelia De Lange Syndrome, Wiedemann-Steiner Syndrome	AD	98,14	144 of 149
KMT2C	Kleefstra Syndrome	AD	98,76	55 of 59
KMT2E	O'donnell-Luria-Rodan Syndrome, Intellectual Disability	AD	99,83	34 of 34
KMT5B	Intellectual Disability	AD	98,35	-
KRAS	Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Noonan Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Encephalocraniocutaneous Lipomatosis, Lynch Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
L1CAM	Corpus Callosum Agenesis, Hydrocephalus, Stenosis Of Aqueduct Of Sylvius, Masa Syndrome	X,XR,G	100	-
LAMB1	Lissencephaly, Cobblestone Lissencephaly	AR	99,97	8 of 9
LAMC3	Cortical Malformations	AR	98,72	22 of 22
LDB1	Nail-Patella Syndrome, Hypotrichosis, Spherocytosis	-	100	1 of 1
LEO1	Fragile X Syndrome, Chiasmal Syndrome, Fibrous Histiocytoma Of Bone, Wilm's Tumor	-	99,84	2 of 2
LHCGR	Hypergonadotropic Hypogonadism, Precocious Puberty	AD,AR	100	75 of 75
LHX1	17q12 Microdeletion Syndrome	-	100	6 of 6
LIG4	Lig4 Syndrome, Dubowitz Syndrome, Omenn Syndrome	AR	99,48	46 of 46
LIMK1	Williams Syndrome	-	100	2 of 2
LMX1B	Nail-Patella Syndrome, 9q33.3q34.11 Microdeletion Syndrome	AD	100	191 of 191
LRP1	Keratosis Pilaris Atrophicans	AR	99,97	30 of 30
LRRC4C	Epileptic Encephalopathy	-	99,59	-
LZTR1	Noonan Syndrome, Schwannomatosis	AD	99,99	136 of 136
MACROD2	Autism Spectrum Disorder, Shizophrenia, Hypogonadotropic Hypogonadism	-	99,93	-
MAGEL2	Prader-Willi Syndrome	AD	99,99	43 of 48
MAN1B1	Intellectual Disability, Congenital Disorder Of Glycosilation	AR	99,97	29 of 30
MAOA	Brunner Syndrome, Monoamine Oxidase A Deficiency	X,XR,G	100	-

MAP1A	Autism Spectrum Disorder	-	99,94	-
MAP1B	Periventricular Nodular Heterotopia	AD	99,28	10 of 12
MAPK1	22q11.2 Microdeletion Syndrome	-	96,91	1 of 1
MBD5	Intellectual Disability, 2q23.1 Microdeletion Syndrome	AD	99,99	33 of 35
MBD6	Wilson Disease, Unverricht-Lundborg Syndrome	-	98,15	7 of 7
MBOAT7	Intellectual Disability	AR	99,08	11 of 12
MCTP2	Monosomy 15q	-	99,95	6 of 6
MECP2	Autism, Encephalopathy, Intellectual Disability, Rett Syndrome, Trisomy Xq28, Psychosis, Macroorchidism Syndrome	X,XR,XD,MU,G	99,81	-
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Blepharophimosis, Intellectual Disability, Fg Syndrome, Marfan Syndrome	X,XR,G	100	-
MED12L	Nizon-Isidor Syndrome	AD	99,94	7 of 7
MED13	Intellectual Developmental Disorder	AD	97,23	17 of 17
MED13L	Intellectual Disability, Cardiac Defects, Developmental Delay, Facial Dysmorphism	AD	100	90 of 92
MEF2C	Intellectual Disability, Stereotypic Movements, Epilepsy, Cerebral Malformations, 5q14.3 Microdeletion Syndrome	AD	99,91	43 of 46
MEIS2	Cleft Palate, Cardiac Defects, Intellectual Disability, 15q14 Microdeletion Syndrome	AD	92	18 of 20
MET	Deafness, Osteofibrous Dysplasia, Renal Cell Carcinoma	AD,AR	99,8	41 of 41
METTL5	Intellectual Developmental Disorder, Microcephaly	AR	99,9	4 of 4
MFRP	Microphthalmia, Retinitis Pigmentosa, Foveoschisis, Nanophthalmos	AR	100	36 of 36
MID1	Opitz Syndrome	X,XR,G	99,95	-
MID2	Intellectual Disability	X,XR,G	99,64	-
MKRN3	Prader-Willi Syndrome, Precocious Puberty	AD,ADWMI	99,98	39 of 41
MKRN3-AS1	Prader-Willi Syndrome	AD	-	-
MKX	Cleft Palate, Acrocallosal Syndrome, Spondylosis, Mitochondrial Complex I Deficiency, Shoulder Impingement Syndrome	-	99,99	1 of 1
MLH1	Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome, Lynch Syndrome	AD,AR	99,94	1079 of 1118
MLH3	Lynch Syndrome	AD	99,98	32 of 32
MLXIPL	Williams-Beuren Syndrome	AD	99,42	-
MSH2	Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99,99	1032 of 1057
MSH6	Mismatch Repair Cancer Syndrome, Lynch Syndrome, Muir-Torre Syndrome	AD,AR	99,28	613 of 641
MTOR	Focal Cortical Dysplasia Of Taylor, Smith-Kingsmore Syndrome, Macrocephaly, Intellectual Disability, Neurodevelopmental Disorder, Small Thorax Syndrome	AD	99,98	39 of 39
MYH10	Lymphangioleiomyomatosis, Hypertelorism, Coloboma Of Macula, Ptosis	-	99,92	9 of 9
MYO5A	Griscelli Syndrome, Neuroectodermal Melanolysosomal Disease	AR	100	10 of 10
MYO9B	Celiac Disease, Ulcerative Colitis, Dermatitis Herpetiformis	-	97,93	5 of 5
MYT1L	Intellectual Disability, 2p25.3 Deletion Syndrome	AD	99,98	30 of 30
NAA15	Intellectual Disability	AD	98,44	39 of 44
NACC1	Neurodevelopmental Disorder, Epilepsy, Cataracts, Delayed Brain Myelination, Feeding Difficulties, Stereotypic Hand Movement	AD	99,99	3 of 3
NAGA	Kanzaki Disease, Schindler Disease, Alpha-N-Acetylgalactosaminidase Deficiency	AR	100	12 of 12
NAV2	Neuroblastoma, Sucrase-Isomaltase Deficiency, Hirschsprung Disease, Attention Deficit-Hyperactivity Disorder	-	99,8	5 of 5
NBEA	Epilepsy, Seizure Disorder, Neurodevelopmental Disorder, Autism Spectrum Disease	-	99,48	27 of 27
NBN	Aplastic Anemia, Leukemia, Nijmegen Breakage Syndrome	AR,MU,P	100	200 of 200
NCKAP1	Autism Spectrum Disorder, Nance-Horan Syndrome, Spinocerebellar Ataxia, Hennekam Syndrome	-	99,97	10 of 10
NCOA1	Endometrial Hyperplasia, Glycogen Storage Disease, Amelogenesis Imperfecta	-	99,61	3 of 3

NCOR1	Rett Syndrome, Endometrial Hyperplasia, Mitochondrial Complex III Deficiency	-	98,92	12 of 12
NDN	Prader-Willi Syndrome	AD	97,41	2 of 2
NDP	Exudative Vitreoretinopathy, Norrie Disease, Coats Disease	X,XR,G	100	-
NECAP1	Epileptic Encephalopathy	AR	99,83	2 of 2
NEGR1	Niemann-Pick Disease, Leptin Deficiency	-	100	1 of 1
NEXMIF	Intellectual Disability	X,XR,XD,G	99,74	-
NF1	Neurofibromatosis, Noonan Syndrome, Watson Syndrome, 17q11.2 Microduplication Syndrome	AD	97,97	3082 of 3166
NFE2L3	Fibrosarcomatous Osteosarcoma	-	99,12	1 of 1
NFIB	Macrocephaly, Intellectual Development	AD	97,92	13 of 14
NFIX	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome	AD	94,42	75 of 81
NHS	Cataract, Nance-Horan Syndrome	X,XD,G	98,45	-
NINL	Trichostrongylosis, Autism Spectrum Disorder, Usher Syndrome, Joubert Syndrome	-	99,92	2 of 2
NIPBL	Cornelia De Lange Syndrome	AD	99,32	409 of 426
NKAP	Intellectual Developmental Disorder	X,XR,G	98,5	-
NLGN1	Autism Spectrum Disorder	AD	98,29	7 of 8
NLGN2	Ritscher-Schinzel Syndrome, Pitt-Hopkins Syndrome, Kaufam Oculocerebrofacial Syndrome, Kagami-Ogata Syndrome, Developmental Disorder	-	96,5	4 of 4
NLGN3	Asperger Syndrome, Autism Spectrum Disorder	X,MU,G	100	-
NLGN4X	Asperger Syndrome, Autism Spectrum Disorder	X,MU,G	99,96	-
NODAL	Heterotaxy, Holoprosencephaly	AD	100	18 of 18
NONO	Intellectual Disability, Macrocephaly	X,XR,G	99,59	-
NOP56	Spinocerebellar Ataxia	AD	99,41	-
NPAP1	Prader-Willi Syndrome	AD	99,82	-
NR2F1	Bosch-Boonstra Optic Atrophy Syndrome, Intellectual Disability	AD	89,78	26 of 31
NR3C2	Hypertension, Pseudohypoaldosteronism	AD	99,99	71 of 72
NR4A2	Parkinson Disease	AD	100	9 of 16
NRXN1	Pitt-Hopkins-Like Syndrome	AR	97,42	33 of 74
NRXN2	Childhood Disintegrative Disorder, Pitt-Hopkins Syndrome, Kaufman Oculocerebrofacial Syndrome, Autism Spectrum Disorder, Developmental Disorder	-	95,53	7 of 8
NRXN3	Childhood Disintegrative Disorder, Autism Spectrum Disorder, Pitt-Hopkins Syndrome	-	99,49	2 of 6
NSD1	Sotos Syndrome, 5q35 Microduplication Syndrome, Weaver Syndrome	AD	99,8	451 of 459
NSDHL	Ck Syndrome, Hemidysplasia	X,XR,XD,G	100	-
NSUN2	Intellectual Disability, Dubowitz Syndrome	AR	99,99	8 of 8
NTNG1	Rett Syndrome, Intellectual Disability	-	99,96	2 of 2
NTRK2	Epileptic Encephalopathy, Developmental Delay, West Syndrome	AD	100	9 of 9
NUAK1	Omphalocele, Squamous Cell Carcinoma	-	99,94	2 of 2
NUP155	Atrial Fibrillation	AR	99,91	2 of 3
NUS1	Congenital Disorder Of Glycosylation, Intellectual Disability, Seizures, Epileptic Encephalopathy	AD,AR	99,62	22 of 23
OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	-
ODC1	Global Developmental Delay, Alopecia, Macrocephaly, Facial Dysmorphism, Structural Brain Anomalies	AD	100	7 of 7
OPHN1	Intellectual Disability, Cerebellar Hypoplasia	X,XR,G	100	-
OTUD7A	Learning Disability, Chromosome 15q13.3 Deletion Syndrome, Schizophrenia, Epilepsy	-	84,69	1 of 2

OTX2	Microphthalmia, Pituitary Hormone Deficiency, Holoprosencephaly, Situs Inversus, Septo-Optic Dysplasia Spectrum	AD	100	56 of 58
OXTR	Prosopagnosia, Adenomyosis, Borderline Personality Disorder, Alexithymia	-	98,39	1 of 1
P2RX5	Cystinosis, Chronic Fatigue Syndrome	-	100	1 of 1
P4HA2	Myopia	AD	99,98	11 of 11
PACS1	Intellectual Disability, Craniofacial Dysmorphism, Cryptorchidism	AD	97,98	3 of 3
PAFAH1B1	Lissencephaly, 17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	AD	99,95	90 of 92
PAH	Phenylketonuria	AR	100	964 of 969
PAK2	Human Immunodeficiency Virus	-	99,98	1 of 1
PAK3	Intellectual Disability	X,XR,G	99,96	-
PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, Pallidal Degeneration, Neurodegeneration	AR	98,92	177 of 182
PARD3B	Hypertension, Schizophrenia, Amyotrophic Lateral Sclerosis	-	99,94	2 of 2
PARS2	Epileptic Encephalopathy	AR	100	7 of 7
PAX5	Leukemia, Gray Zone Lymphoma	-	100	8 of 8
PCDH19	Epilepsy, Intellectual Disability, Dravet Syndrome	X,G	99,99	-
PCDH9	Chromosomal 9p Deletion Syndrome, Neuropathy	-	99,96	-
PCGF2	Turnpenny-Fry Syndrome	AD	89,88	2 of 2
PCNT	Microcephalic Osteodysplastic Primordial Dwarfism, Seckel Syndrome	AR	99,92	103 of 105
PDE10A	Limb And Orofacial Dyskinesia, Striatal Degeneration, Chorea	AD,AR	100	8 of 8
PDE4D	Acrodysostosis, Haploinsufficiency Syndrome	AD	98,73	37 of 39
PER2	Advanced Sleep-Phase Syndrome	AD	100	7 of 7
PHB	Breast Cancer	AD	100	1 of 1
PHF12	Tarsal Tunnel Syndrome, Neuropathy, Failure Of Tooth Eruption	-	95,38	2 of 2
PHF2	Dissociative Disorder, Culler-Jones Syndrome, Autism Spectrum Disorder	-	99,95	2 of 2
PHF21A	Intellectual Developmental Disorder, Behavioral Abnormalities, Craniofacial Dysmorphism, Seizures, Potocki-Shaffer Syndrome	AD	99,83	9 of 10
PHF3	Retinitis Pigmentosa, Cone-Rod Dystrophy	-	99,45	8 of 9
PHF6	Borjeson-Forssman-Lehmann Syndrome	X,XR,G	99,93	-
PHIP	Developmental Delay, Intellectual Disability, Dysmorphic Features	AD	98,74	51 of 52
PHRF1	Lupus Erythematosus, Cartilage-Hair Hypoplasia	-	99,94	5 of 5
PIEZO2	Arthrogryposis, Gordon Syndrome, Marden-Walker Syndrome, Oculomotor Limitation, Electroretinal Anomalies Syndrome	AD,AR	96,93	37 of 37
PIGL	Zunich Neuroectodermal Syndrome, Chime Syndrome, Hyperphosphatasia, Intellectual Disability	AR	86	11 of 13
PIK3CA	Overgrowth, Vascular Malformations, Keratosis, Macrocephaly, Capillary Malformation, Hemihyperplasia, Lynch Syndrome, Meningioma	AD	99,58	54 of 58
PIP5K1B	Fredreich Ataxia	-	99,83	-
PLCB1	Epileptic Encephalopathy, West Syndrome	AR	99,92	4 of 6
PLXNA4	Polycystic Kidney Disease, Policystic Liver Disease	-	99,99	5 of 5
PLXND1	Moebius Syndrome	-	98,44	6 of 6
PMS1	Lynch Syndrome	AD	99,92	32 of 33
PMS2	Colorectal Cancer, Mismatch Repair Cancer Syndrome, Lynch Syndrome	AD,AR	97,17	264 of 285
PNKP	Ataxia-Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AR	100	36 of 36
POGZ	White-Sutton Syndrome, Intellectual Disability, Microcephaly, Strabismus, Behavioral Abnormalities	AD	99,97	85 of 85
POLA1	Pigmentary Disorder, Van Esch-O'driscoll Syndrome, Intellectual Disability	X,XR,G	99,26	-

POMGNT1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99,91	82 of 83
PON1	Amyotrophic Lateral Sclerosis	-	100	8 of 8
PON3	Amyotrophic Lateral Sclerosis	-	100	3 of 3
PPM1D	Breast Cancer, Intellectual Developmental Disorder, High Pain Threshold	AD	97,82	76 of 79
PPP1R12A	Genitourinary And Brain Malformation Syndrome	AD	99,48	1 of 1
PPP1R9B	Limbic Encephalitis, Ventricular Tachycardia, Schizophrenia, Alzheimer Disease	-	99,5	-
PPP2R5D	Intellectual Disability, Macrocephaly, Hypotonia, Behavioral Abnormalities	AD	100	11 of 11
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, Intellectual Development, Epileptic Encephalopathy	AD	99,98	16 of 16
PQBP1	Renpenning Syndrome, Intellectual Disability	X,XR,G	99,99	-
PRDM16	Left Ventricular Noncompaction, 1p36 Deletion Syndrome, Cardiomyopathy	AD	98,81	20 of 20
PREX1	Gastric Tubular Adenocarcinoma	-	98	3 of 3
PRICKLE1	Epilepsy, Unverricht-Lundborg Disease	AR	98,41	23 of 23
PRICKLE2	Sensory Ataxic Neuropathy, Epilepsy,	-	94,92	6 of 6
PRKAR1A	Acrodysostosis, Carney Complex, Myxoma, Pigmented Nodular Adrenocortical Disease, Acute Promyelocytic Leukemia	AD	95,93	165 of 171
PRKCB	Diabetic Macular Edema, Hyperglycemia	-	96,24	3 of 4
PRKCG	Spinocerebellar Ataxia	AD	100	52 of 52
PRKD2	Polycystic Kidney Disease, Policystic Liver Disease, Mitochondrial Complex I Defficiency	-	97,12	2 of 2
PRKN	Lung Cancer, Parkinson Disease	AD,AR	100	-
PRNP	Creutzfeldt-Jakob Disease, Insomnia, Gerstmann-Straussler Disease, Huntington Disease, Kuru, Spongiform Encephalopathy, Neuropsychiatric Features, Alzheimer Disease	AD	100	69 of 69
PRODH	Hyperprolinemia, Schizophrenia	AD,AR	98,57	5 of 5
PROKR2	Hypogonadotropic Hypogonadism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD	100	64 of 64
PRR12	Coloboma Of Iris, Cat Eye Syndrome, Autism, Mirror Movements,	-	98,69	3 of 3
PSMD12	Stankiewicz-Isidor Syndrome, 17q24.2 Microdeletion Syndrome, Intellectual Disability	AD	97,93	3 of 4
PTCH1	Basal Cell Nevus Syndrome, Holoprosencephaly, Gorlin Syndrome, Monosomy 9q22.3	AD	98,89	498 of 502
PTCHD1	Autism, Intellectual Disability	X,XR,G	99,98	-
PTEN	Cowden Disease, Macrocephaly, Autism Spectrum Disorder, Meningioma, Bannayan-Riley-Ruvalcaba Syndrome, Lhermitte-Duclos Disease, Proteus Syndrome, Segmental Outgrowth, Arteriovenous Malformation	AD	99,97	609 of 629
PTK7	Panic Disorder, Social Phobia, Agoraphobia, Anxiety, Neural Tube Defects	-	99,99	14 of 14
PTPN11	Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome	AD	100	150 of 151
PWAR1	Prader-Willi Syndrome	AD	-	-
PWRN1	Prader-Willi Syndrome	AD	-	-
PYHIN1	Scleroderma	-	98,67	2 of 3
QRICH1	Ververi-Brady Syndrome	AD	99,94	16 of 16
RAB2A	Sezary Syndrome, Warburg Micro Disease	-	98,62	2 of 2
RAB39B	Intellectual Disability, Parkinson Disease	X,XR,G	100	-
RAB43	Cone Rod Dystrophy	-	100	3 of 3
RAD21	Cornelia De Lange Syndrome, Mungan Syndrome	AD,AR	99,8	16 of 17
RAI1	Smith-Magenis Syndrome, 17p11.2 Microduplication Syndrome, Contiguous Gene Duplication Syndrome	AD	99,91	50 of 53
RALGAPB	Septooptic Dysplasia, Tubulinopathies	-	99,98	25 of 25
RANBP17	Spinocerebellar Ataxia, Parkinson Disease	-	99,97	1 of 1

RBFOX1	Epilepsy, Spinocerebellar Ataxia, Developmental Coordination Disorder, Autism Spectrum Disorder	-	97,99	4 of 5
RBM27	Mucocutaneous Leishmaniasis	-	93,84	2 of 2
RELN	Epilepsy, Lissencephaly	AD,AR	100	70 of 70
RERE	Neurodevelopmental Disorder, Anomalies Of The Brain, 1p36 Deletion Syndrome	AD	92,43	21 of 21
REV3L	Moebius Syndrome	-	99,08	7 of 7
RFC2	Williams Syndrome	-	100	3 of 3
RFX3	Histrionic Personality Disorder, Visceral Heterotaxy, Ciliary Diskinesia, Chromosome 9p Deletion Syndrome	-	99,96	4 of 4
RIC1	Catifa Syndrome	AR	99,9	-
RIMS1	Cone Rod Dystrophy	AD	98,2	24 of 24
RLIM	Intellectual Disability	X,XR,G	99,52	-
ROBO2	Vesicoureteral Reflux	AD	99,78	20 of 20
RORA	Intellectual Developmental Disorder, Epilepsy, Cerebellar Ataxia	AD	99,94	12 of 12
RORB	Epilepsy	AD	99,98	4 of 4
RPL10	Intellectual Disability, Cerebellar Hypoplasia, Spondyloepiphyseal Dysplasia, Microcephaly, Growth Retardation, Prognathism, Cryptorchidism	X,XR,G	100	-
RPS20	Colorectal Cancer	-	99,97	1 of 1
RPS6KA3	Coffin-Lowry Syndrome, Intellectual Disability	X,XD,G	99,95	-
RREB1	22q11.2 Deletion Syndrome	-	99,92	8 of 8
RSRC1	Intellectual Developmental Disorder, Intellectual Disability	AR	99,8	2 of 2
SAE1	Dermatomyositis	-	99,97	-
SATB1	Glass Syndrome, Breast Disease	-	99,96	-
SATB2	Chromosome 2q32-Q33 Deletion Syndrome	AD	99,87	97 of 124
SBF1	Charcot-Marie-Tooth Disease	AR	99,94	19 of 19
SCN1A	Epileptic Encephalopathy, Febrile Convulsions, Migraine, Dravet Syndrome, Hemiplegic Migraine, Febrile Seizures, Lennox-Gastaut Syndrome	AD	99,8	1776 of 1797
SCN2A	Epileptic Encephalopathy, Episodic Ataxia, Seizures, Dravet Syndrome, Febrile Seizures, West Syndrome	AD	100	351 of 351
SCN3A	Epileptic Encephalopathy	AD	99,98	18 of 18
SCN8A	Cognitive Impairment, Cerebellar Ataxia, Epileptic Encephalopathy, Myoclonus, Seizures, Convulsions, Choreoathetosis	AD	97,85	156 of 172
SCN9A	Erythermalgia, Generalized Epilepsy, Febrile Seizures, Indifference To Pain, Neuropathy, Paroxysmal Extreme Pain Disorder, Dravet Syndrome	AD,AR	96,25	126 of 137
SDHB	Carney-Stratakis Syndrome, Paragangliomas, Pheochromocytoma, Cowden Syndrome, Succinate-Coq Reductase Deficiency	AD	100	261 of 264
SDHC	Carney-Stratakis Syndrome, Paragangliomas, Cowden Syndrome, Pheochromocytoma	AD	99,95	62 of 63
SDHD	Carney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paragangliomas, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase Deficiency	AD,AR	99,98	164 of 166
SEC23B	Anemia, Cowden Syndrome	AD,AR	100	119 of 127
SEC24C	22q11.2 Deletion Syndrome	-	99,98	-
SEMA3E	Charge Syndrome, Hypogonadotropic Hypogonadism	AD,AR	99,81	6 of 7
SEMA4A	Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR	99,94	15 of 15
SEMA5A	Monosomy 5p	-	100	7 of 7
SET	Intellectual Disability	AD	98,84	8 of 8
SETBP1	Intellectual Disability, Expressive Aphasia, Facial Dysmorphism, Schinzel-Giedion Syndrome	AD	98,61	43 of 43
SETD2	Luscan-Lumish Syndrome, Sotos Syndrome	AD	99,83	19 of 19
SETD5	Intellectual Disability, Cornelia De Lange Syndrome, Facial Dysmorphism	AD	99,77	37 of 37

SGSH	Mucopolysaccharidosis	AR	97,7	151 of 151
SH2B1	16p11.2 Microdeletion Syndrome, Insulin Resistance	-	99,98	25 of 25
SH3KBP1	Agammaglobulinemia	X,XR,G	99,55	-
SHANK1	Schizophrenia, Autism Spectrum Disorder, Phelan-Mcdermid Syndrome, Trichomegaly	-	89,73	10 of 11
SHANK2	Autism Spectrum Disorder, Developmental Disorder, Phelan-Mcdermid Syndrome	-	99,91	64 of 64
SHANK3	Phelan-Mcdermid Syndrome, Schizophrenia, Monosomy 22q13.3	AD,MU,P	96,67	-
SHH	Holoprosencephaly, Microphthalmia, Schizencephaly, Hypoplastic Tibiae, Radial Hemimelia	AD	99,48	161 of 184
SHOX	Langer Mesomelic Dysplasia, Leri-Weill Dyschondrosteosis, Short Stature	AD,AR,X,G	99,98	-
SIM1	6q16 Microdeletion Syndrome, Prader-Willi Syndrome	-	99,64	39 of 40
SIN3A	Chromosome 15q24 Deletion Syndrome, Intellectual Disability	AD	99,94	18 of 18
SIX3	Holoprosencephaly, Schizencephaly	AD	99,79	79 of 80
SKI	Shprintzen-Goldberg Craniosynostosis Syndrome, 1p36 Deletion Syndrome, Shprintzen-Goldberg Syndrome	AD	99,66	39 of 39
SLC12A5	Epileptic Encephalopathy	AD,AR	100	19 of 19
SLC13A5	Epileptic Encephalopathy, Amelocerebrohypohidrotic Syndrome	AR	95,92	24 of 24
SLC16A2	Allan-Herndon-Dudley Syndrome	X,XR,G	99,94	-
SLC1A2	Epileptic Encephalopathy	AD	100	7 of 7
SLC2A1	Choreoathetosis, Spasticity, Epilepsy, Glucose Transport Defect, Cryohydrocytosis, Episodic Ataxia	AD,AR	99,99	301 of 304
SLC35A3	Arthrogryposis, Intellectual Disability, Seizures, Autism Spectrum Disorder, Epilepsy	AR	99,94	5 of 5
SLC35B1	Dicarboxylic Aminoaciduria, Hydranencephaly	-	99,49	-
SLC35C1	Congenital Disorder Of Glycosylation	AR	99,73	8 of 8
SLC38A10	Brittle Bone Syndrome	-	99,78	-
SLC6A1	Myoclonic-Atonic Epilepsy, Myoclonic-Astastic Epilepsy	AD	100	55 of 55
SLC6A3	Dystonia, Parkinson Disease	AR	100	31 of 31
SLC6A4	Obsessive-Compulsive Disorder	AD	99,95	-
SLC6A8	Creatine Deficiency Syndrome	X,XR,G	99,87	-
SLC7A3	Aphakia, Vestibular Nystagmus	-	99,87	-
SLC7A5	Phenylketonuria, Lysinuric Protein Intolerance, Maple Syrup Urine Disease	-	99,34	3 of 3
SLC9A6	Intellectual Disability, Christianson Syndrome	X,XD,G	98,87	-
SLC9A7	Intellectual Developmental Disorder, Intellectual Disability	X,XR,G	97,1	-
SLC9A9	Autism Spectrum Disorder, Attention-Deficit Hiperactivity Disorder, Pervasive Developmental Disorder, Christianson Syndrome	-	100	11 of 11
SLITRK1	Gilles De La Tourette Syndrome, Trichotillomania	AD,MU	100	10 of 12
SLITRK5	Trichotillomania, Body Dysmorphic Disorder, Obsessive-Compulsive Disorder, Impulse Control Disorder, Tourette Syndrome	-	100	10 of 10
SMAD4	Myhre Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia	AD	99,56	136 of 136
SMARCA4	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome	AD	100	68 of 69
SMARCC2	Coffin-Siris Syndrome	AD	99,49	16 of 16
SMC1A	Cornelia De Lange Syndrome, Holoprosencephaly, Wiedemann-Steiner Syndrome	X,XR,XD,G	100	-
SMC3	Cornelia De Lange Syndrome	AD	100	30 of 30
SMG6	Miller-Dieker Lissencephaly Syndrome	-	98,84	1 of 1
SMPD1	Niemann-Pick Disease	AR	99,98	258 of 258
SNORD115-1	Prader-Willi Syndrome	AD	-	-
SNORD116-1	Prader-Willi Syndrome	AD	-	-

SNRPN	Autism, Prader-Willi Syndrome	AD,MU	100	2 of 2
SNX5	Fanconi Anemia, Encephalitis, Parkinson Disease	-	79,68	2 of 2
SON	Zttk Syndrome, Brain Malformations, Musculoskeletal Abnormalities, Facial Dysmorphism, Intellectual Disability	AD	99,27	30 of 32
SOX2	Microphthalmia, Esophageal Atresia, Septo-Optic Dysplasia Spectrum	AD	99,91	78 of 78
SOX3	Intellectual Disability, Panhypopituitarism, Septo-Optic Dysplasia Spectrum	X,G	92,88	-
SOX5	Lamb-Shaffer Syndrome, Developmental And Speech Delay	AD	99,95	9 of 9
SPARCL1	Entropion	-	98,01	1 of 1
SPAST	Spastic Paraplegia	AD	99,98	616 of 655
SPEN	Mullegama-Klein-Martinez Syndrome, Wolfan Syndrome	-	99,9	8 of 8
SPG7	Spastic Paraplegia, Primary Lateral Sclerosis	AD,AR	99,94	125 of 126
SPRED1	Legius Syndrome	AD	100	84 of 84
SRCAP	Floating-Harbor Syndrome	AD	99,98	53 of 53
SRPR	Myoclonus Epilepsy, Hematopoietic System Disease, Schopf-Schulz-Passarge Syndrome, Ollier Disease	-	99,97	-
SRPX2	Rolandic Epilepsy, Intellectual Disability, Speech Dyspraxia, Perisylvian Polymicrogyria	AD	100	-
SRSF11	Alzheimer Disease, Neurodegenerative Disease, Bipolar Disorder, Intellectual Disability	-	99,84	2 of 2
ST7	Autism Spectrum Disorder	-	99,91	2 of 2
ST8SIA2	Amelogenesis Imperfecta, Visual Cortex Disease, Dysentery	-	99,94	1 of 1
STAG2	Holoprosencephaly, Neurodevelopmental Disorder, Craniofacial Abnormalities, Xq25 Microduplication Syndrome	X,XR,G	99,09	-
STK3	Intellectual Disability	-	99,86	0 of 2
STS	Ichthyosis	X,XR,G	100	-
STXBP1	Epileptic Encephalopathy, 9q33.3q34.11 Microdeletion Syndrome, Rett Syndrome, Dravet Syndrome, West Syndrome	AD	100	209 of 215
STXBP5	Von Willebrand Disease, Epileptic Encephalopathy	-	99,78	4 of 4
SUPT16H	Alpha-Thalassemia, Intellectual Disability, Hiperinsulinemic Hypoglycemia Robinow Syndrome	-	99,81	5 of 5
SVBP	Neurodevelopmental Disorder, Ataxia, Hypotonia, Microcephaly, Intellectual Disability	AR	100	-
SYN2	Schizophrenia	AD	-	-
SYNGAP1	Intellectual Disability, Epileptic Encephalopathy	AD	99,46	168 of 171
SYNJ1	Epileptic Encephalopathy, Parkinson Disease	AR	99,81	30 of 32
SYP	Intellectual Disability	X,XR,G	99,98	-
SZT2	Epileptic Encephalopathy	AR	99,98	39 of 39
TAF1	Dystonia, Intellectual Disability, Parkinson Disease, Global Development Delay, Facial Dysmorphism	X,XR,G	99,74	-
TAF6	Alazami-Yuan Syndrome	AR	100	5 of 5
TANC2	Intellectual Developmental Disorder, Autism Spectrum Disorder, Language Delay, Seizures, Intellectual Disability	AD	97,81	21 of 21
TAOK1	Intellectual Disability, Autism Spectrum Disorder	-	99,94	8 of 8
TAOK2	Intellectual Disability, Autism Spectrum Disorder	-	99,75	5 of 5
TBC1D20	Warburg Micro Syndrome	AR	99,94	6 of 6
TBC1D24	Deafness, Doors Syndrome, Epileptic Encephalopathy, Myoclonic Epilepsy, Dystonia, Rolandic Epilepsy	AD,AR	100	80 of 80
TBC1D31	Branchiootorenal Syndrome	-	99,8	2 of 2
TBCK	Hypotonia, Psychomotor Retardation, Intellectual Disability	AR	99,95	15 of 15
TBL1X	Hypothyroidism	X,G	98,73	-
TBL1XR1	Intellectual Disability, Pierpont Syndrome, Promyelocytic Leukemia	AD	99,78	23 of 23

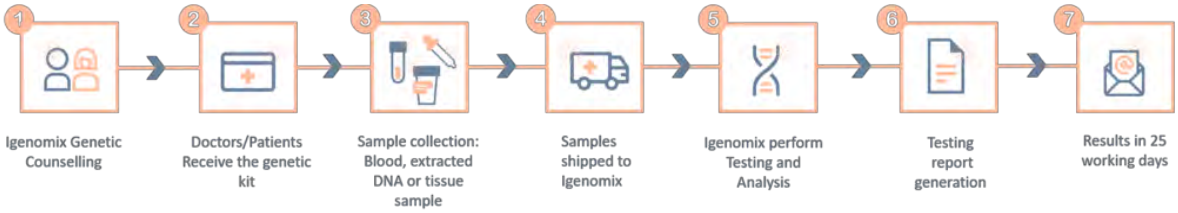
TBL2	Williams Syndrome	-	96,14	-
TBR1	Intellectual Developmental Disorder, Autism Spectrum Disorder, Speech Delay, 2q24 Microdeletion Syndrome	AD	99,04	13 of 13
TBX1	Conotruncal Heart Malformations, Digeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome	AD,AR	88,7	35 of 42
TCF12	Craniosynostosis, Brachycephaly, Plagiocephaly	AD	99,98	73 of 76
TCF20	Developmental Delay, Intellectual Impairment, Behavioral Abnormalities	AD	100	73 of 73
TCF4	Corneal Dystrophy, Pitt-Hopkins Syndrome, Primary Sclerosing Cholangitis	AD	98,91	124 of 124
TCF7L2	Diabetes Mellitus	AD	99,79	5 of 5
TDGF1	Holoprosencephaly	-	100	3 of 3
TEK	Glaucoma, Venous Malformations	AD,AR	100	35 of 35
TERF2	Werner Syndrome, Nijmegen Breakage Syndrome, Bloom Syndrome, Congestive Splenomegaly, Hoyeraal Hreidarsson Syndrome	-	97,59	8 of 8
TET2	Myelodysplastic Syndrome, Sideroblastic Anemia, Systemic Mastocytosis, Thrombocythemia, Polycythemia Vera, Myelofibrosis, Refractory Anemia	-	99,96	15 of 15
TET3	Beck-Fahrner Syndrome	AD,AR	97,53	1 of 1
TGFBR2	Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome	AD	99,9	165 of 166
TGIF1	Holoprosencephaly	AD	99,94	23 of 23
THRB	Thyroid Hormone Resistance	AD,AR	99,94	178 of 178
TIMM8A	Mohr-Tranebjaerg Syndrome	X,XR,G	100	-
TKT	Developmental Delay, Congenital Heart Defects, Transketolase Deficiency	AR	99	6 of 6
TLK2	Intellectual Disability	AD	96,98	39 of 39
TM9SF4	Melanoma	-	100	1 of 1
TMCO1	Cerebrofaciothoracic Dysplasia	AR	88	5 of 5
TMLHE	Autism	X,XR,G	81,62	-
TNRC6B	Epilepsy, Autism Spectrum Disorder, Murray Valley Encephalitis	-	99,87	21 of 21
TRAF7	Cardiac, Facial, And Digital Anomalies, Developmental Delay, Meningioma	AD	100	5 of 5
TRAK1	Epileptic Encephalopathy	AR	99,28	7 of 7
TRAPPC14	Microcephaly	AR	-	-
TRAPPC4	Neurodevelopmental Disorder, Epilepsy, Spasticity, Brain Atrophy, Intellectual Disability	AR	100	-
TRAPPC9	Intellectual Disability, Brain Malformations, Facial Dysmorphism	AR	100	17 of 18
TRIM23	Cerebellar Degeneration, Mulibrey Nanism	-	99,95	1 of 1
TRIO	Intellectual Developmental Disorder, Macrocephaly, Intellectual Disability, Micrognathia, Behavioral Abnormalities	AD	96,84	35 of 36
TRIP12	Intellectual Disability	AD	99,92	31 of 31
TRMT1	Intellectual Developmental Disorder, Intellectual Disability	AR	99,97	5 of 5
TRPC6	Focal Segmental Glomerulosclerosis	AD	99,92	52 of 55
TRPM1	Night Blindness	AR	99,3	87 of 87
TRRAP	Deafness, Developmental Delay, Dysmorphic Facies, Autism	AD	99,98	46 of 46
TSC1	Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	99,86	390 of 406
TSC2	Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	100	1157 of 1159
TSHZ3	Hydronephrosis, Athabaskan Brainstem Dysgenesis Syndrome, Anomalies Of Kidney And Urinary Tract	-	94,08	2 of 2
TSPAN7	Intellectual Disability	X,XR,G	99,97	-
TUBB2B	Cortical Dysplasia, Brain Malformations, Dysequilibrium Syndrome, Polymicrogyria	AD	84,28	29 of 38
UBA5	Epileptic Encephalopathy, Spinocerebellar Ataxia	AR	99,98	19 of 19

UBE3A	Angelman Syndrome, 15q11q13 Microduplication Syndrome	AD	99,98	208 of 211
UBE3C	Neuronopathy, Coenzime Q10 Deficiency, Limb Girdle Muscular Dystrophy, Parkinson Disease,	-	99,22	3 of 3
UBN2	Autism Spectrum Disorder	-	96,08	5 of 5
UBR1	Johanson-Blizzard Syndrome	AR	100	72 of 72
UBR5	Autism Spectrum Disorder, Williams-Beuren Disorder, Myoclonic Epilepsy, Johanson-Blizzard Syndrome	-	99,98	7 of 7
UFD1	22q11.2 Deletion Syndrome	-	99,98	-
UNC79	Esotropia	-	99,97	2 of 2
UPF3B	Intellectual Disability	X,XR,G	98,75	-
USF3	Cowden Syndrome	-	99,61	-
USP27X	Intellectual Disability	X,XR,G	99,84	-
USP45	Leber Congenital Amaurosis	AR	99,08	4 of 5
USP7	Chromosome 16p13.2 Deletion Syndrome	AD	99,98	18 of 18
USP9X	Intellectual Disability, Facial Dysmorphism, Choanal Atresia	X,XR,XD,G	98,61	-
VAMP2	Neurodevelopmental Disorder, Hypotonia, Autism Spectrum Disorder	AD	99,62	5 of 5
VEZF1	Spherocystosis, Arthrogryposis, Diamond-Blackfan Anemia,	-	98,88	-
VIL1	Milker's Nodule, Chemical Colitis, Space Motion Sickness	-	99,92	3 of 3
VPS13B	Cohen Syndrome	AR	99,98	182 of 190
WAC	Desanto-Shinawi Syndrome, Facial Dysmorphism, Developmental Delay, Behavioral Abnormalities	AD	98,98	35 of 35
WASF1	Neurodevelopmental Disorder, Absent Language, Seizures	AD	97,03	3 of 3
WDFY3	Microcephaly, Intellectual Disability	AD	99,95	60 of 60
WDFY4	Lupus Erythematosus	-	99,98	5 of 5
WFS1	Cataract, Deafness, Diabetes Mellitus, Wolfram Syndrome	AD,AR	99,97	390 of 395
WWOX	Epileptic Encephalopathy, Spinocerebellar Ataxia, Intellectual Disability	AR	99,94	44 of 44
YWHAG	Epileptic Encephalopathy	AD	99,94	5 of 5
YY1	Gabriele-De Vries Syndrome	AD	99,89	13 of 13
ZBTB20	Ossified Ear Cartilages, Muscle Wasting, Intellectual Disability, Cataract, Myopathy	AD	97,04	32 of 33
ZC3H4	Silicosis, Cornelia De Lange Syndrome, Specific Language Impairment,	-	95,92	3 of 3
ZDHHC9	Intellectual Disability	X,G	100	-
ZEB2	Mowat-Wilson Syndrome	AD	98,95	253 of 254
ZIC2	Holoprosencephaly	AD	84,47	86 of 112
ZMIZ1	Neurodevelopmental Disorder, Dysmorphic Facies, Distal Skeletal Anomalies, Intellectual Disability	AD	98,87	13 of 13
ZMYND11	Intellectual Disability	AD	99,83	16 of 16
ZMYND8	Lymphoma	-	98,54	2 of 2
ZNF292	Intellectual Developmental Disorder, Disease Of Mental Health, Microcephaly, Alacrima, Achalasia, Intellectual Disability, Autism Spectrum Disorder	-	99,95	32 of 32
ZNF407	Microcephaly, Radioulnar Synostosis, Intellectual Disability, Homocarnosinosis	-	99,96	6 of 6
ZNF41	Intellectual Disability	-	99,98	-
ZNF462	Weiss-Kruszka Syndrome	AD	100	21 of 21
ZNF507	Seckel Syndrome	-	99,72	-
ZNF711	Intellectual Disability	X,G	99,83	-
ZNF804A	Schizophrenia, Bipolar Disorder, Epileptic Encephalopathy	-	99,42	2 of 2
ZNF81	Intellectual Disability	-	99,56	-

ZNHIT6	Retinitis Pigmentosa, Sotos Syndrome	-	96,73	-
ZSWIM6	Acromelic Frontonasal Dysostosis, Neurodevelopmental Disorder, Movement Abnormalities, Autism Spectrum Disorder	AD	91,16	2 of 2

*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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