



Syndrome Coverage

Our Human [ACCUArray™](#) offers unparalleled coverage with over 19,000 clones providing near-tiling resolution through the entire genome. The list below contains many known syndromes and recently discovered genetic conditions which are covered on our [ACCUArray™](#).

Disease/Syndrome	Location	Genes
10q22.3-q23.31 Microdeletion	10q22.3-q23.31	Multiple
12q14.1-q15 Microdeletion	12q14.3	LEMD3, GRIP1
12q24.21-q24.23 Microduplication	12q24.21-q24.33	Multiple
14q22-q23 Microdeletion	14q22-q23	Multiple
15q24.1-q24.3 Microdeletion	15q24.1-q24.3	Multiple
16p11.2-p12.2 Microdeletion/ Microduplication	16p11.2-p12.2	Multiple
16p13.1 Microdeletion/Microduplication	16p13.1	Multiple
16p13.3 Microdeletion/Severe Rubinstein-Taybi	16p13.3	CREBBP, DNASE1
17q21.3 Microdeletion	17q21.3	MAPT
1q21.1 Microdeletion	1q21.1	Multiple
1q41-q42 Microdeletion/Fryns	1q41	DISP1
22q11.2 Duplication	22q11.2	TBX1
22q13.3 Microdeletion	22q13.3	ARSA, SHANK3
2p15-p16.1 Microdeletion	2p15-p16.1	Multiple
2q32.2-q33 Microdeletion	2q33.1	SATB2
3q29 Microdeletion	3q29	Multiple
5q Syndrome/ 5q Deletion Syndrome	5q33.1	MEGF1, G3BP
6p25.3 Microdeletion	6p25.3	Multiple
7q11.23 Duplication	7q11.23	Multiple
8p23.1 Microdeletion/Microduplication	8p23.1	GATA4
9q22.32-q22.33 Microdeletion	9q22.33	TGFBR1
9q34 Microdeletion	9q34.3	EHMT1
Adrenal hypoplasia congenita	Xp21.2	NROB1 , DAX1
Adult-onset autosomal dominant leukodystrophy	5q23.2	LMNB1
Alagille	20p12.2	JAG1
Albright hereditary osteodystrophy	2q37.3	Multiple
Alpha thalassemia mental retardation (OS)	16p13.3	HBA1, HBA2
Alzheimers Disease	21q21.3	APP
AMME Complex/ Alport Syndrome	Xq22.3	COL4A5, COL4A6
Angelman Syndrome (AS) (duplications or deletions)	15q11.2-12	UBE3A, D15S10
Aniridia II	11p13	PAX6
Atrial septal defect (ASD)	5q35.1	NKX2-5
Autistic Features	15q11.2-q13	Multiple

Azoospermia / Oligozoospermia	Yq11.223	Multiple
Bartter 1	15q21.1	SLC12A1
Bartter 2	11q24.3	KCNJ1
Bartter 3 (classic)	1p36.13	CLCNKB
Bartter 4 (infantile with sensorineural deafness)	1p32.3,p36.13	BSND, CLCNKA
Down (Downs) syndrome critical region (DSCR)	21q22.13	Multiple
Duane Retraction Syndrome 1 / DURS1/ DRS/ DUS	8q13.1	Multiple
Duchenne Muscular Dystrophy (DMD)	Xp21.2-p21.1	DMD,
Familial adenomatous polyposis (FAP)	5q22.2	APC
Feingold	2p24.3	MYCN
FG 5	Xq22.3	MID2
FMR1 Microdeletion	Xq27.3	FMR1
Gitelman	16q13	SLC12A3
Glycerol kinase deficiency	Xp21.2	GK
Glycerol Kinase Deficiency / Hyperglycerolemia	Xp21.2	GK
Greig cephalopolysyndactyly	7p14.1	GLI3
Holoprosencephaly 1	21q22.3	TMEM1
Holoprosencephaly 3	7q36.3	SHH
Holoprosencephaly 5	13q32.3	ZIC2
Holoprosencephaly 6	2q37.1-q37.3	HPE6
Holoprosencephaly 8	14q13.1-q13.2	Multiple
Holoprosencephaly and preaxial polydactyly	5q35.1	FBXW11
Holt-Oram	12q24.1	TBX5
Hypodontia, Autosomal Dominant (HYD1)	14q12-q13	PAX9
Hypoparathyroidism, sensorineural deafness	10p14	GATA3
Hypotonia-Cyctinura Syndrome	2p21	SLC3A1, PREPL
Iris Hypoplasia / Rieger anomaly (RA)	6p25	FOXC1
Jacobsen 11q terminal deletion disorder	11q24.3-q25	Multiple
Joubert 3	6q23.3	AHI1
Joubert 4/ Nephronophthisis 1	2q13	NPHP1
Joubert 5	12q21.32	CEP290
Joubert Syndrome 4	2q13	NPHP1
Kallmann's Syndrome 1 / KAL1	Xp22.31	KAL1
Klinefelters, XXY	+X, +Y	
Langer-Giedion	8q23.3-q24.11	EXT1, TRPS1
Leri-Weill dyschondrosteosis / Langers	Xpt22.33-p22.32	SHOX, SHOXY
Li-Fraumeni 1	17p13.1	TP53
Lissencephaly	17p13.3	LIS1
Lissencephaly with cerebellar hypoplasia	7q22.1	RELN
Loeys-Dietz (LDS)	9q22.33	TGFBR1
Loeys-Dietz (LDS)/Marfan 2 (MFS2)	3p24.1	TGFBR2
Lowe	Xq25	OCRL
Lymphoproliferative Syndrome, X-Linked / XLP / XLPD	Xq25	SH2D1a

Madelung Deformity / Leri-Weill dyschond -rosteosis	Xp22.33	SHOX, DXS6796
Marfan	15q21.1	FBN1
Medulloblastoma/ MDB	10q24-q26	DMBT1, SUFU
Microphthalmia 3	3q26.33	SOX2
Microphthalmia 7 with linear skin defects	Xp22.2	HCCS
Miller-Dieker/Lissencephaly 1	17p13.3	LIS1
Monosomy 1p36 Microdeletion	1p36	Multiple
Monosomy 9p Syndrome	9p22.3-p23	Multiple
Mowat-Wilson	2q22.3	ZFXH1B
Multiple Exostoses, type II/ EXT2	11p11.2	EXT2
Nablus mask-like facial	8q21.3-q22.1	Multiple
Nail-patella	9q33.3	LMX1B
Nephronophthisis 1 (NPHP1) / Nephronophthisis	2q13	NPHP1
Neurofibromatosis 1/MR	17q11.2	NF1
Neurofibromatosis 2	22q12.2	NF2
Neuropathy / HNPP	17p12	PMP22
NFIA Haploinsufficiency	1p31.3	NFIA
Noonan 1 Syndrome / NS1	12q24.13	PTPN11
Noonan 4	2p22.1	SOS1
Oculocutaneous Albinism Type 1	11q14.3	TYR
Oculocutaneous Albinism Type 2	15q13.1	OCA2
Okhiro	20q13.2	SALL4
Opitz	Xp22.2	MID1
Ornithine transcarbamylase deficiency	Xp11.4	OTC
Orofaciodigital 1	Xp22.2	OFD1
Pallister-Killian	12p	KRAS2, LDHB
Parietal Foramina 1 (PFM1) / Foramina Parietalia	5q35.2	MSX2
Parietal Foramina 2 (PRM2)	11p11.2	ALX4,
Pelizaeus-Merzbacher	Xq22.2	PLP1, PLP
Polycystic kidney disease (OS) / PKD1	16p13.3	PKD1
Potocki-Shaffer Syndrome/ PSS	11p11.2	EXT2, ALX4
Prader-Willi Syndrome(PWS) (duplications or deletions)	1515q11.2	SNRPN, D15S11
Prader-Willi-like phenotype	6q16.3	SIM1
Renal cysts and diabetes (RCAD)	17q12	TCF2
Retinoblastoma/ MR / RB / RB1	13q14.2	RB1
Rett Syndrome/ MECP2 male duplication	Xq28	MECP2
Rieger Syndrome Type 1 / RIEG1 / RIEG / RGS	4q25	PIX2
Rubenstein-Taybi (Rubinstein)	16p13.3	CREBBP
Sacral Agenesis / Currarino Syndrome	7q36.3	HXLB9
Saethre-Chotzen Syndrome (SCS)	7p21.1	TWIST1
Schizencephaly	10q26.11	EMX2
Schizophrenia 4 (SCZD4)	22q11.21	PRODH
Sertoli cell-only syndrome	Yq11.21	USP9Y, DBY

Severe myoclonic epilepsy of infancy (SMEI)	2q24.3	SCN1A
Sex reversal, autosomal dominant 2	9p24.3	Multiple
Short stature, pituitary and cerebellar defects	1q25.2	LHX4
Small cell cancer of the lung / SCLC1 / SCLS / SCCL	3p12.3-p21.31	ACY1, ROBO1
Smith-Lemli-Opitz	11q13.4	DHCR7
Smith-Magenis Syndrome (SMS)	17p11.2	RAI1, FL1
Sotos / Cerebral Gigantism	5q35.3	NSD1
Split-Hand/Foot Malformation 1 (SHFM1)	7q21.3	SHFM1
Split-Hand/Foot Malformation 3 (SHFM3)	10q24.32	FBXW4
Split-Hand/Foot Malformation 4 (SHFM 4)	3q28	TP73L
Split-Hand/Foot Malformation 5 (SHFM 5)	2q31.1	DLX1, DLX2
SRY deletion (OS)	Yp11.31	SRY
Steroid sulfatase deficiency	Xp22.31	STS
Stickler I	12q13.11	COL2A1
Swyer Syndrome / SRY / XY Gonadal Dysgenesis	Yp11.31	SRY
Synpolydactyly / SPD / Syndactyly II	2q31.1	HOXD
Thyroid Hormone Resistance	3p24.2	THRB
Townes-Brocks	16q12.1	SALL1
Trichorhinophalangeal 1	8q23.3	TRPS1
Trichorhinophalangeal syndrome / Langer-Giedion	8q24.11	TRPS1, EXT1
Tuberous sclerosis 2 (OS)	16p13.3	TSC2
Turner Syndrome, X	-X	
Ulnar-Mammary	12q24.1	TBX3
Van Buchem Disease (VBCH)	17q12-21	SOST
Van der Woude Syndrome	1q32.2	IRF6
Vascular endothelial growth factor disorders	6p21.1	VEGFA
Velocardiofacial	22q11.2	TBX1,
Von Hippel-Lindau Syndrome / VHL	3p23	VHL,
Waardenburg syndrome IIA	3p13	MITF
WAGR Syndrome	11p13	PAX6, WT1
Williams-Beuren	7q11.23	ELN, LIMK1
Wilms Tumor 1 / WT1 / Nephroblastoma	11p13	WT1
Wolf-Hirschhorn (WHS)	4p16.3	WHSCR, WHSCR2
X-linked agammaglobulinemia	Xq22.1	BTK
X-linked heterotaxy	Xq26.3	ZIC3
X-linked infantile spasms	Xp22.13	CDKL5
X-linked juvenile retinoschisis	Xp22.13	RS1
X-linked lissencephaly	Xq23	DCX
X-linked lymphoproliferative syndrome (XLP)	Xq25	SH2D1A
X-linked mental retardation 21	Xp21.3	IL1RAPL1
X-linked mental retardation 30	Xq23	PAK3
X-linked mental retardation 54/	Xp21.3	ARX
X-linked mental retardation	Xq27.1	SOX3

Xp11.3 Microdeletion	Xp11.3	RP2, ZNF674
XY Sex-reversal, +/- adrenal failure	9q33.3	NR5A1