ECS_AR_X-linked panel

versie	V1 (124 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABCB7	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive
ABCD1	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ACSL4	300157	Mental retardation, X-linked 63, 300387 (3), X-linked dominant
ALAS2	301300	Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked
AMER1	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
AR	313700	Androgen insensitivity, 300068 (3), X-linked recessive; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
ARSL (ARSE)	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ARX	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
ATP7A	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BRWD3	300553	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
BTK	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive

CACNA1F	300110	Aland Island eye disease, 300600 (3), X-linked; Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked
CASK	300172	FG syndrome 4, 300422 (3); Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; Mental retardation, with or without nystagmus, 300422 (3)
CCNQ	300708	STAR syndrome, 300707 (3), X-linked dominant
CD40LG	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
CDKL5	300203	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
CFP	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
CHM	300390	Choroideremia, 303100 (3), X-linked dominant
CLCN5	300008	Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive; Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive
COL4A5	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
COX7B	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CUL4B	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
CYBB	300481	Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive; Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive
DCX	300121	Lissencephaly, X-linked, 300067 (3), X-linked; Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLG3	300189	Mental retardation, X-linked 90, 300850 (3), X-linked recessive
DMD	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
EBP	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
EDA	300451	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive; Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant
EFNB1	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
F8	300841	Hemophilia A, 306700 (3), X-linked recessive
F9	300746	{Deep venous thrombosis, protection against}, 300807 (3); Hemophilia B, 306900 (3), X-linked recessive; Thrombophilia, X-

		linked, due to factor IX defect, 300807 (3); {Warfarin sensitivity}, 122700 (3), Autosomal dominant
FANCB	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FGD1	300546	Aarskog-Scott syndrome, 305400 (3), X-linked recessive; Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive
FHL1	300163	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive
FLNA	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; Congenital short bowel syndrome, 300048 (3), X-linked recessive; ?FG syndrome 2, 300321 (3), X-linked; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant
FMR1	309550	Fragile X syndrome, 300624 (3), X-linked dominant; Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
FOXP3	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
FRMPD4	300838	Mental retardation, X-linked 104, 300983 (3), X-linked
G6PD	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
GATA1	305371	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GK	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLA	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked

GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GRIA3	305915	Mental retardation, X-linked 94, 300699 (3), X-linked recessive
HCCS	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3), X-linked recessive
HDAC8	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HPRT1	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HSD17B10	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HUWE1	300697	Mental retardation, X-linked syndromic, Turner type, 309590 (3), X-linked
IDS	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IGBP1	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
IKBKG	300248	Ectodermal dysplasia and immunodeficiency 1, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
IL1RAPL1	300206	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
IL2RG	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive
IQSEC2	300522	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIF4A	300521	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive
L1CAM	308840	CRASH syndrome, 303350 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; MASA syndrome, 303350 (3), X-linked recessive
LAMP2	309060	Danon disease, 300257 (3), X-linked dominant
LAS1L	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
MBTPS2	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive

MECP2	300005	{Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, atypical, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MED12	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MTM1	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive
NAA10	300013	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NDP	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
NEXMIF	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
NHS	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
NR0B1	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
NSDHL	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
OCRL	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
OFD1	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
OTC	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
PAK3	300142	Mental retardation, X-linked 30/47, 300558 (3), X-linked recessive
PCDH19	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PGK1	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PHF6	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHF8	300560	Mental retardation syndrome, X-linked, Siderius type, 300263 (3), X-linked recessive
PHKA2	300798	Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive

PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POU3F4	300039	Deafness, X-linked 2, 304400 (3), X-linked recessive
PQBP1	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRPS1	311850	Arts syndrome, 301835 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive
RBM10	300080	TARP syndrome, 311900 (3), X-linked recessive
RP2	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RPGR	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
RPL10	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive
RPS6KA3	300075	Coffin-Lowry syndrome, 303600 (3), X-linked dominant; Mental retardation, X-linked 19, 300844 (3), X-linked dominant
RS1	300839	Retinoschisis, 312700 (3), X-linked recessive
SH2D1A	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SHROOM4	300579	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
SLC16A2	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC35A2	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism
SLC6A8	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC9A6	300231	Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
SMS	300105	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive
SOX3	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
STS	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
SYP	313475	Mental retardation, X-linked 96, 300802 (3), X-linked recessive

TAZ	300394	Barth syndrome, 302060 (3), X-linked recessive
TBX22	300307	?Abruzzo-Erickson syndrome, 302905 (3), X-linked; Cleft palate with ankyloglossia, 303400 (3), X-linked
TIMM8A	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TSPAN7	300096	Mental retardation, X-linked 58, 300210 (3), X-linked recessive
UBA1	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
UPF3B	300298	Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive
USP9X	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
WAS	300392	Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
ZIC3	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Sept 30, 2019

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.