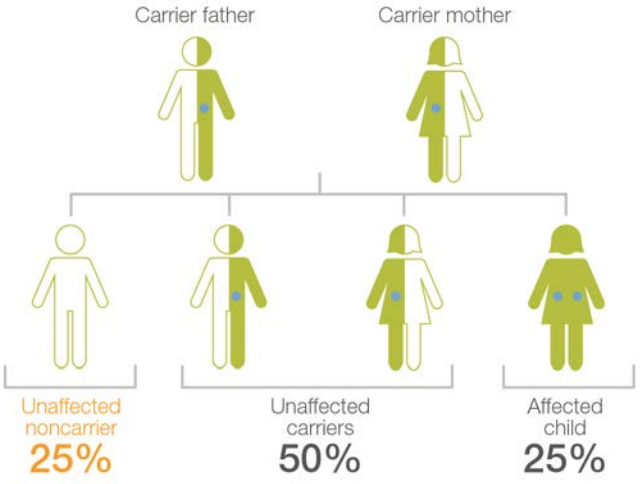
genescreen®

About GeneScreen® - Carrier Screening Test

GeneScreen® is an advanced **carrier screening test** that identify couples who are at risk of passing inherited disorders to their children.

Expectant parents, or couples planning a family, may be at risk for passing on severe genetic diseases to their offspring. If both parents are carriers for an autosomal recessive or X-linked condition they have a 1 in 4, or 25%, chance of having an affected child.



The **GeneScreen® Carrier Screen** provides a closer look at genes, to see if the couple is at risk of passing a hereditary genetic disorder to their offspring.

GeneScreen® Carrier Screening Test allows for a comprehensive care and enables patients to make more informed reproductive decisions. Offering **GeneScreen®** to a patient before pregnancy allows her to gain knowledge about her reproductive health early.

Carrier testing with comprehensive coverage

Thanks to the introduction of the latest technologies, including **Next Generation Sequencing** (**NGS**), it is now practical and affordable to test patients for a broad range of genetic disorders that are individually rare but collectively common. This enables healthcare providers to offer a thorough risk assessment to all patients regardless of family history or ethnicity.

Using NGS technique, **GeneScreen® Expanded** - Carrier Screening Test screens for several of the most commonly requested disorders, including those recommended by the ACMG & ACOG and disorders specif ic for individuals of Ashkenazi Jewish descent, testing **925 genes** and **1500+** recessive and X-linked **genetic disorders**. This provides patients more opportunities to identify potential hereditary risks.

The **GeneScreen®** Test provides a comprehensive study of genes included on the panel, with a high detection rate for each condition, identifying a high number of couples at reproductive risk (Haque et al., 2016).

Carrier screen with unmatched detection of serious disorders

The true goal of carrier screening is to detect at-risk couples of serious, prevalent, and clinically-actionable diseases. That's why the **GeneScreen®** test has been methodically designed by the genetic experts to maximize detection rates for the diseases that matter the most.

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The **GeneScreen®** test screens for the most clinically relevant and impactful genetic conditions that typically affect health in infancy or childhood. The disorders included in the **GeneScreen®** test panel have been carefully selected for preconception and prenatal carrier testing. Each condition is selected based on carrier rate, clinical severity, and availability of treatment options. These disorders can cause serious health problems, intellectual disability, or a shorter life. Testing can be performed before conception, or at any time during pregnancy.

GeneScreen® Carrier Screening Test screens severe diseases according to the guidelines of the European Society of Human Genetics (ESHG) (Henneman et al., 2016), including those recommended by prestigious international societies, such as the American College of Medical Genetics and Genomics (ACMG) and the American College of Obstetricians and Gynecologists (ACOG)(Edwards et al., 2015)

Knowing Enables Informed Decisions

Knowledge of carrier status by **GeneScreen®** Test before conception allows at-risk couples to make informed reproductive choices. There are a number of potential choices to consider:
If both individuals test positive as carriers of the same recessive condition, they can review options such as *in vitro* fertilization (IVF) and preimplantation genetic diagnosis (PGD) to improve the chances of having an unaffected child. PGD is available at GENOMA for any gene included in the **GeneScreen®** panel.
Other couples may decide to become pregnant naturally, undergoing to prenatal diagnosis. For these patients, GENOMA may offer an extensive catalog of prenatal tests, including all genes available in the **GeneScreen®** panel.
The couple can also be prepared and obtain early intervention for children with certain disorders to improve outcomes. Individuals identified as carriers can also inform family members of their potential risk.

GeneScreen®: Indication for testing

GeneScreen® Test is intended to be used as a family planning tool, allowing patients to be tested individually or with their reproductive partner for their risk of having children with various genetic conditions. This test is intended to identify couples with high reproductive risk. Thus, concurrent testing of both prospective parents is highly recommended.

GeneScreen® is intended for patients who meet any of the following criteria:

- Individuals with a family history of a genetic disease, who are therefore at higher risk of being carriers for those diseases
- Individual belonging to certain ethnicities with high risk of being carriers of hereditary recessive disorders.
- For patients who are pursuing pregnancy with assisted reproductive technologies.
- Couples planning to start a family or to extend it, and willing to know if they are carriers of monogenic recessive diseases that can be transmitted to their offspring, in order to make more informed reproductive decisions.
- Couples requiring gamete donation, in order to select the most appropriate donor for each recipient (i.e. a donor that doesn't carry the same mutation as the member of the couple who will provide the gametes), minimizing the reproductive risk.
- -Couples who are already pregnant and who wish to know more about the genetic health of their pregnancy;

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- Gamete banks or IVF clinics to analyze every egg or sperm donor, allowing also the recipients analysis to avoid high reproductive risk pregnancies.
- Anyone who wants to know if they are carrier of any condition included in the panel.

Supported by genetic counseling, carrier screening programs have been successful in reducing the incidence of inherited diseases. The American College of Medical Genetics (ACMG) and American College of Obstetrics and Gynecology (ACOG) recommend that couples of reproductive age be offered carrier screening before conception (Edwards et al., 2015).

GeneScreen®: The Testing Process

The DNA is first isolated from the peripheral blood and then **amplified by PCR.** Through a state-of-the-art technological process, named **massively parallel sequencing** (**MPS**), which uses ***Next Generation Sequencing*** (**NGS**) techniques with **ILLUMINA** sequencing instruments, **925 genes** are completely sequenced (whole exons sequencing, including adjacent intronic regions, ± 5 nucleotides) (Table 1) at high read depth. The resulting genetic sequences are analysed via an **advanced bioinformatics analysis**, to assess the presence of potential mutations in the genes under investigation. **GeneScreen®** test, unlike other carrier screening tests using targeted sequencing, performs **full-exon sequencing** of all the genes included on the panel, which allows a more comprehensive analysis of each gene and related diseases. Gene dosage analysis by Multiplex Ligation-dependent Probe Amplification (MLPA) of the SMN1 gene was performed for SMA carrier screening. Fluorescent PCR was used for Fragile-X carrier screen, to detect the (CGG)n repeat expansions in the promoter region of the FMR-1 gene.

Results of the GeneScreen® test

Positive Result: One or more disease-causing change(s) was detected in the gene(s) included in **GeneScreen®** panel identifying the patient as a **carrier**. Mutations can be classified as:

- **Known pathogenic:** clinical relevant mutations causing well-established syndromes;
- **Likely pathogenic:** variants that are likely clinical relevant and may cause well-established syndromes.

Being a carrier is relatively common. Carriers do not generally show any symptoms. A patient with a positive test result should be referred for genetic counseling and further evaluation. The patient's reproductive partner and at-risk family members may also be tested. When both parents are carrier of the same genetic disease, genetic counseling is recommended to the couple where various reproductive choices will be discussed, including:

- **Invasive or Non Invasive Prenatal diagnostic test during pregnancy**
- **Preimplantation Genetic Diagnosis (PGD)**
- **Egg or Sperm donation.**

Unclear Result: A change was detected in the gene/region of interest but currently, there is insufficient information in the medical literature to know whether is a disease-causing change or a normal variation in the population. This kind of changes are defined **"Variants of uncertain clinical significance (VOUS)".**

Classification follows the recommendations of the international reference guidelines. [12]

Negative Result: No known disease-causing change was not detected in the genes included in **GeneScreen®** panel. A single test cannot always detect all possible genetic changes. Hence, negative results do not completely rule out the presence of a disease-causing change in the investigated genes.

Bioinformatics Analysis For Variant Interpretation

Analysis of NGS data is a complex process, imposing challenging requirements both in terms of computing

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resources and software. **GeneScreen®** Test uses powerful custom-built bioinformatic solutions to support variant analysis that enables fast, reliable and highly accurate results. When a variant is detected during the sequencing process, its pathogenicity will be investigated in accordance with ACMG and AMP guidelines[8], using a sophisticated software. A team of board-certified geneticists provide expert interpretation and clearly explained reports.

Parameters used to report the genetic variations

The test analyses only the genes listed in Table 1. Only variants classified as "**known pathogenic**", "**Likely pathogenic"** and **Variants of uncertain clinical significance (VOUS),** in accordance with the relevant scientific literature and the current classification in the ClinVar – NCBI, dbSNP – NCBI, and other NCBI resources, Human Gene Mutation Database (HGMD), updated on the date of the sample collection, will be reported. Moreover, in compliance with the indications of the American College of Medical Genetics (ACMG), only mutations with a Minor Allele Frequency (MAF) <5% (1000 Genomes Project) are considered as pathogenic or possibly pathogenic; this measurement refers to the frequency in which the less common allele is present in the general population.

Target Coverage

Target Coverage is the average number of sequencing reads for each nucleotide base of the gene. Variations with a read depth (i.e. number of reads) lower than 30X are not detected by the bioinformatics analysis algorithm.

Accuracy of the GeneScreen® test

Current DNA sequencing techniques are **more than 99%** accurate. While results of this testing are highly accurate, a negative result significantly reduces but does not eliminate the chance of being a carrier. The results of this testing, including the benefits and limitations, should be discussed with patients.

Limitation of the GeneScreen® test

This test analyses only genetic diseases and genes listed in Table 1. The test does not detect other genetic diseases or genes that were not specifically targeted.
Moreover, the test cannot detect:

- mutations located in the intronic regions beyond ± 5 nucleotides from the breakpoints;
- deletions, inversions, or duplications of more than 20 bps;
- germline mosaicism (i.e. mutations occurring only in the gametes)

A **"NEGATIVE" - No mutations** result for the genes analyzed does not exclude the possibility that mutations are present in a region of the genome that was not explored during the analysis. Some regions of our DNA may not be sequenced or have a lower coverage than the limitations set by GENOMA Group experts to guarantee an accurate examination of gene variations. These regions, therefore, are not included in the analysis if they do not meet the requested qualitative standards. In some cases the result of genome testing may reveal DNA variations or mutations with an unknown or unclassifiable clinical significance with the current medical and scientific knowledge. Moreover, the detection of gene variations does not always imply that the person will develop a certain pathology or the severity of the related symptoms, nor when this person may have the disease. The value of some gene variations detected through this test, therefore, may not be classified with the current medical and scientific knowledge.
The interpretation of genetic variations is based upon the most updated knowledge available upon examination. Such interpretation may change in the future, when new scientific and medical information on the structure of the genome are acquired and may affect the evaluation of the genetic variations

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themselves.
Some pathologies may be caused or regulated by more than one variation in the DNA, in one or more genes. Some of these variations may not be identified or validated yet by the scientific community and, therefore, may not be classified as pathogenic variations at the time of analysis.
For a correct interpretation of results, we need to have accurate information on the health of the patient and any pathology in the clinical history of the couple and their relatives. This information allows our geneticists to have a better interpretation of genetic results.
The intrinsic limitation of the NGS methodology is the lack of coverage uniformity of each examined genetic region. Quantity and quality of the DNA extracted from prenatal samples is one of the potential causes of such lack of uniformity, which may lead to the lack of detection of gene mutations. Due to this limitation, NGS tests may not detect specific genetic mutations in the selected genes.

Genetic Counseling

Genetic counseling is essential for any patient found to be a mutation-carrier for a genetic disorder. Genoma will provide a genetic counseling session for those patients that screen positive, and this service is included in the cost of the test. It aids the patient in medical comprehension and enhances patient satisfaction by providing access to experts who are skilled at explaining genetic risks in terms patients can understand.

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Table 1: GeneScreen® Expanded - List of genes screened

AAAS	ATP7B	COL6A3	ESPN	GNPTAB	KCNV2	NBN	PEX6	SELENON	TGM1
ABCA12	ATP8B1	COL7A1	ESRRB	GNPTG	KDM5C	NDP	PEX7	SEMA4A	TH
ABCA3	ATR	COL9A1	ETFA	GNRHR	KIAA2022	NDRG1	PFKM	SEPSECS	THRA
ABCA4	ATRX	COL9A2	ETFB	GNS	KIF7	NDUFA1	PGK1	SERPINA1	THRB
ABCB11	AUH	COQ2	ETFDH	GORAB	L1CAM	NDUFA7	PGM1	SETX	TIMM8A
ABCB4	B4GALT1	COQ8A	ETHE1	GP1BA	LAMA2	NDUFAF2	PHF8	SFTPB	TK2
ABCB7	B9D2	COQ9	EVC	GP1BB	LAMA3	NDUFAF4	PHGDH	SFTPC	TLR3
ABCC2	BBS1	COX10	EVC2	GP9	LAMB2	NDUFAF5	PHKG2	SGCA	TMC1
ABCC6	BBS10	COX15	EXOSC3	GPC3	LAMB3	NDUFS3	PHYH	SGCB	TMEM216
ABCC8	BBS12	COX6B1	EYS	GPR143	LAMC2	NDUFS4	PKHD1	SGCD	TMEM67
ABCD1	BBS2	CPS1	F11	GPR179	LAMP2	NDUFS5	PKLR	SGCG	TMIE
ABCD4	BCHE	CPT1A	F2	GRHPR	LARGE1	NDUFS6	PLA2G6	SGSH	TMPRSS3
ACAD8	BCKDHA	CPT2	F5	GRIA3	LBR	NDUFS7	PLCE1	SH2D1A	TNFRSF11B
ACAD9	BCKDHB	CRB1	F8	GRIK2	LCA5	NDUFS8	PLEC	SH3TC2	TNNT1
ACADL	BCOR	CRLF1	F9	GRM6	LDHA	NDUFV1	PLEKHG5	SHROOM4	TPO
ACADM	BCS1L	CRTAP	FAH	GRXCR1	LDLR	NEB	PLG	SIL1	TPP1
ACADS	BEST1	CRX	FAM126A	GSS	LDLRAP1	NEFL	PLOD1	SIX6	TPRN
ACADSB	BLM	CSTB	FAM161A	GTF2H5	LHCGR	NEU1	PLP1	SLC12A1	TRAPPC9
ACADVL	BLOC1S6	CTH	FAM20C	GUCY2D	LHFPL5	NEUROG3	PMM2	SLC12A3	TRDN
ACAT1	BRCA2	CTNS	FANCA	GUSB	LHX3	NHEJ1	PMP22	SLC12A6	TREX1
ACE	BRIP1	CTSC	FANCB	HADH	LIFR	NHLRC1	PNPO	SLC16A2	TRIM32
ACOX1	BRWD3	CTSD	FANCC	HADHA	LIG4	NHP2	POLG	SLC17A5	TRIM37
ACSF3	BSCL2	CTSK	FANCD2	HADHB	LIPA	NHS	POLR1C	SLC19A2	TRIOBP
ACSL4	BSND	CUL4B	FANCE	HAL	LIPH	NKX2-1	POMGNT1	SLC22A5	TRMU
ACTN4	BTD	CYBA	FANCG	HAMP	LMBRD1	NKX2-5	POMT1	SLC24A1	TSEN54
ADA	BTK	CYBB	FANCI	HAX1	LMNA	NLGN3	POMT2	SLC25A13	TSFM
ADAMTS13	C3	CYP11A1	FANCL	HBA1	LOXHD1	NLGN4X	POR	SLC25A15	TSHB
ADAMTS2	CA2	CYP11B1	FANCM	HBA2	LPL	NLRP7	POU1F1	SLC25A20	TSHR
ADAMTSL2	CANT1	CYP11B2	FAS	HBB	LRAT	NMNAT1	POU3F4	SLC25A22	TSPAN7
ADGRG1	CAPN3	CYP17A1	FASLG	HCCS	LRP2	NOP10	PPT1	SLC26A2	TSPYL1
ADGRV1	CASK	CYP19A1	FASTKD2	HESX1	LRP5	NPC1	PQBP1	SLC26A3	TTC37
ADK	CASP10	CYP1B1	FBLN5	HEXA	LRPPRC	NPC2	PRCD	SLC26A4	TTN

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AFF2	CASQ2	CYP21A2	FERMT3	HEXB	LRTOMT	NPHP1	PRF1	SLC26A5	TTPA
AGA	CBS	CYP27A1	FGA	HFE	LYST	NPHP3	PRKRA	SLC35A1	TUBA1A
AGL	CC2D2A	CYP27B1	FGB	HFE2	MAGT1	NPHP4	PRODH	SLC35C1	TUFM
AGPS	CCDC103	CYP4V2	FGD1	HGD	MAK	NPHS1	PROM1	SLC35D1	TULP1
AGT	CCDC39	CYP7B1	FGD4	HGF	MAN2B1	NPHS2	PROP1	SLC37A4	TUSC3
AGTR1	CD19	D2HGDH	FH	HGSNAT	MARVELD 2	NR0B1	PRPS1	SLC39A4	TWNK
AGTR2	CD247	DBT	FHL1	HIBCH	MAT1A	NR2E3	PRSS12	SLC3A1	TYK2
AGXT	CD2AP	DCLRE1 C	FIG4	HLCS	MATN3	NR5A1	PRX	SLC45A2	TYMP
AHCY	CD320	DCX	FKRP	HMGCL	MBTPS2	NSD1	PSAP	SLC46A1	TYR
AHI1	CD3D	DDB2	FKTN	HMOX1	MCCC1	NSDHL	PSAT1	SLC4A11	TYRP1
AIPL1	CD3E	DDC	FLNA	HOGA1	MCCC2	NSUN2	PTEN	SLC5A5	UBA1
AIRE	CD3G	DFNB59	FLVCR1	HP	MCEE	NTRK1	PTH1R	SLC6A19	UBE2A
ALAS2	CD40LG	DGUOK	FMR1	HPD	MCOLN1	NUP62	PTS	SLC6A8	UBE3A
ALDH3A2	CDH23	DHCR24	FOLR1	HPRT1	MCPH1	NXF5	PUS1	SLC7A7	UBR1
ALDH4A1	CDH3	DHCR7	FOXG1	HPS1	MECP2	NYX	PYGM	SLC7A9	UGT1A1
ALDH5A1	CDHR1	DHDDS	FOXN1	HPS3	MED12	OAT	QDPR	SLC9A6	UNC13D
ALDH7A1	CDK5RAP 2	DKC1	FOXP3	HSD11B2	MED17	OCA2	RAB23	SLX4	UNC93B1
ALDOA	CDKL5	DLD	FRAS1	HSD17B1 0	MED25	OCRL	RAB27A	SMARCAL 1	UPF3B
ALDOB	CENPJ	DLG3	FREM2	HSD17B3	MEFV	OFD1	RAB39B	SMN1	UQCRB
ALG1	CEP152	DLL3	FTCD	HSD17B4	MERTK	OPA3	RAB3GAP 1	SMN2	UQCRQ
ALG12	CEP290	DMD	FTSJ1	HSD3B2	MESP2	OPHN1	RAB3GAP 2	SMPD1	UROS
ALG2	CERKL	DMP1	FUCA1	HSPD1	MFRP	ORAI1	RAD51C	SMS	USH1C
ALG3	CFH	DNAH5	FXN	HSPG2	MFSD8	OSTM1	RAG1	SNAI2	USH1G
ALG6	CFP	DNAI1	G6PC	HTRA1	MGAT2	OTC	RAG2	SNAP29	USH2A
ALG8	CFTR	DNAI2	G6PC3	HUWE1	MID1	OTOA	RAPSN	SOX3	USP9X
ALG9	CHM	DNAJC19	G6PD	HYAL1	MKKS	OTOF	RARS2	SP110	VDR
ALMS1	CHRNA1	DNAL1	GAA	HYLS1	MKS1	OXCT1	RAX	SPG11	VIPAS39
ALPL	CHRND	DNMT3B	GALC	ICOS	MLC1	P3H1	RDH12	SPG20	VLDLR
ALS2	CHRNE	DOCK8	GALE	IDH3B	MLYCD	PAH	RDX	SPG7	VPS13A
AMACR	CHRNG	DOK7	GALK1	IDS	MMAA	PAK3	RELN	SRD5A2	VPS13B
AMPD1	CHST6	DOLK	GALNS	IDUA	MMAB	PALB2	REN	SRD5A3	VPS33B
AMT	CIITA	DPAGT1	GALNT3	IFNGR1	MMACHC	PANK2	RFT1	SRPX2	VRK1
ANO5	CLCN1	DPM1	GALT	IFNGR2	MMADHC	PAX3	RGR	ST3GAL3	VSX2

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ANTXR2	CLCN5	DPYD	GAMT	IFT80	MOCS1	PAX6	RHO	ST3GAL5	VWF
AP1S1	CLCN7	DSP	GAN	IGBP1	MOCS2	PAX8	RLBP1	STAR	WAS
AP1S2	CLDN1	DUOX2	GBA	IGF1	MOGS	PC	RMRP	STAT1	WDR62
AP3B1	CLDN14	DUOXA2	GBE1	IGHMBP 2	MPDU1	PCBD1	RNASEH2 A	STIL	WFS1
APTX	CLDN19	DYNC2H 1	GCDH	IKBKAP	MPI	PCCA	RNASEH2 B	STIM1	WHRN
AQP2	CLN3	DYSF	GCH1	IKBKG	MPL	PCCB	RNASEH2 C	STRA6	WISP3
AR	CLN5	EDA	GCSH	IL12B	MPV17	PCDH15	RP2	STRC	WNT10A
ARG1	CLN6	EDAR	GDAP1	IL12RB1	MPZ	PCDH19	RPE65	STX11	WNT3
ARHGEF6	CLN8	EDN3	GDF5	IL1RAPL 1	MRE11	PDE6A	RPGR	STXBP2	WNT7A
ARHGEF9	CLRN1	EDNRB	GDI1	IL1RN	MRPS16	PDE6B	RPGRIP1L	SUCLA2	WRN
ARL13B	CNGA1	EFEMP2	GFM1	IL2RA	MRPS22	PDE6C	RPL10	SUCLG1	XIAP
ARL6	CNGA3	EFNB1	GHRHR	IL2RG	MTHFR	PDE6G	RPS6KA3	SUMF1	XPA
ARSA	CNGB1	EGR2	GJA1	IMPDH1	MTM1	PDHA1	RRM2B	SUOX	XPC
ARSB	CNGB3	EIF2AK3	GJB1	IMPG2	MTMR2	PDHB	RS1	SURF1	ZDHHC9
ARSE	COG1	EIF2B5	GJB2	INPP5E	MTR	PDHX	RYR1	SYN1	ZEB2
ARSF	COG7	ELK1	GJB3	INSR	MTRR	PDP1	SACS	SYP	ZFYVE26
ARX	COG8	EMD	GJB6	INVS	MTTP	PDSS1	SAG	TAF1	ZIC3
ASL	COL11A1	ENO3	GJC2	IQCB1	MUT	PDSS2	SAMD9	TAT	ZMPSTE24
ASNS	COL17A1	ENPP1	GK	IQSEC2	MVK	PDX1	SAMHD1	TAZ	ZNF41
ASPA	COL18A1	EPM2A	GLA	ISCU	MYD88	PDZD7	SBDS	TBCE	ZNF469
ASPM	COL1A1	ERBB3	GLB1	ITGA6	MYO15A	PEPD	SBF2	TCAP	ZNF674
ASS1	COL1A2	ERCC2	GLDC	ITGB4	MYO3A	PEX1	SC5D	TCF4	ZNF711
ATIC	COL2A1	ERCC3	GLE1	IVD	MYO5A	PEX10	SCN2A	TCIRG1	ZNF81
ATM	COL4A3	ERCC4	GLIS3	IYD	MYO6	PEX12	SCNN1A	TCN2	
ATP6AP2	COL4A4	ERCC5	GM2A	JAK3	MYO7A	PEX13	SCNN1B	TECTA	
ATP6V0A2	COL4A5	ERCC6	GNAS	KCNJ1	NAGA	PEX2	SCNN1G	TERT	
ATP6V1B1	COL6A1	ERCC8	GNE	KCNJ11	NAGLU	PEX26	SCO1	TFR2	
ATP7A	COL6A2	ESCO2	GNMT	KCNJ13	NAGS	PEX5	SCO2	TG	

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