CarrierCheck (146 genes) Carrier Screening Test

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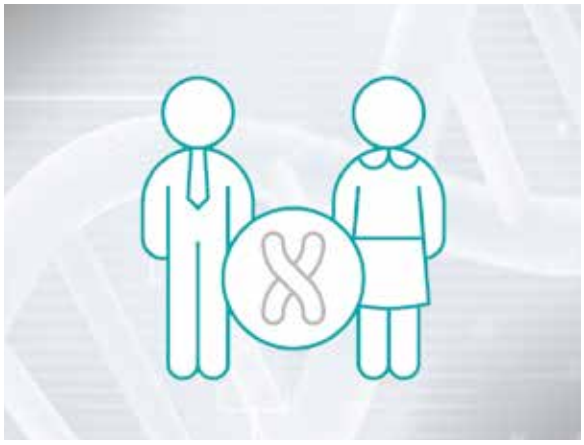
REPRODUCTIVE GENETICS COMPANY

Who is a carrier?

A carrier is an individual who has a mutation in one of the alleles of a gene associated with a genetic disease.



> *"CarrierCheck enables genetic matching of gamete donors to their recipients"*

What is the CarrierCheck test?

Expanded preconceptional carrier test enables the detection of couples at risk for single gene diseases. It allows couples to make the right reproductive decision and reduces the risk of having a affected child. It specifically screens autosomal recessive and X-linked recessive inherited disorders. Next-Generation Sequencing (NGS) empowered by powerful bioinformatic tools enable simultaneous screening of hundreds of diseases with a single universal method.

> *"Carrier Tests can aid to prevent genetic disorders"*

Why CarrierCheck test?

- Comprehensive screening capacity with expanded gene panel - 146 genes
- High variant detection sensitivity,
- Simultaneous detecion of CNVs, SNPs with a single NGS based test
- Special analysis algorithms for efficient diagnosis of challenging gene regions –pseudogenes and homologous genes - SMN1, HBA1/2, CYP21A2, DMD, CFTR, GBA
- Additional test for FMR1 detection
- Exon level CNV detection for critical diseases - DMD, CFTR
- Fast and reliable results with exclusive analysis tool developed by Franklin by Genoox.

> *"Next generation risk reduction for consanguineous couples"*

Who are the candidates of carrier screening?

All couples who want to reduce the risk of having a child with genetic disease can be referred.

- Consanguineous individuals
- People at high risk for a particular disease, based on their ethnic background
- Before IVF applications and donor cycles

Novel reporting module:

> *"Personalized genetic counselling with residual carrier risk"*

- Artificial intelligance supported variant classification
- Ethnic specific residual carrier risk reporting
- Tracking of test process
- Custom tailored genetic counselling: To inform patients about the test-limitations and couple spesific risks
- Genetic matching is enabled via duo-analysis of gamete donors and their recipients
- Patient specific variant reporting algorithm

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GENE	CONDITION
ABCA4	Cone-rod dystrophy 3; Fundus flavimaculatus; Retinitis pigmentosa 19; Stargardt disease 1; Retinal dystrophy
ABCB11	Progressive familial intrahepatic cholestasis, type II
ABCD1	Adrenoleukodystrophy; Adrenomyeloneuropathy, adult
ACADM	Medium chain Acyl-CoA dehydrogenase deficiency
ACADVL	Very long chain Acyl-CoA dehydrogenase deficiency
ACAT1	Beta-ketothiolase deficiency (Alpha-methylacetoacetic aciduria)
ADA	Severe combined immunodeficiency due to ADA deficiency
AGA	Aspartylglycosaminuria
AGL	Glycogen storage disease, type III (a&b)
AGXT	Primary hyperoxaluria, type I
AIRE	Polyglandular autoimmune syndrome, type I
ALDH3A2	Sjögren-Larsson syndrome
ALDOB	Hereditary fructose intolerance
ALPL	Hypophosphatasia, autosomal recessive
ARG1	Argininemia
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type VI
ASL	Argininosuccinic aciduria
ASPA	Canavan disease
ASS1	Citrullinemia, type I
ATM	Ataxia-telangiectasia
ATP7B	Wilson disease
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS4	Bardet-Biedl syndrome 4
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BTD	Biotinidase deficiency
CAPN3	Limb-girdle muscular dystrophy, type 2A
CBS	Homocystinuria, CBS-related
CEP290	Leber congenital amaurosis 10; Joubert syndrome 5; Meckel syndrome 4; Senior-Loken syndrome 6
CFTR	Cystic fibrosis;
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related; Northern epilepsy
CLRN1	Usher syndrome, type IIIA
CNGA3	Achromatopsia 2
COL4A3	Alport syndrome
COL4A4	Alport syndrome 2
CPS1	Carbamoylphosphate synthetase I deficiency
CPT1A	Carnitine palmitoyltransferase IA deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CTNS	Cystinosis, CTNS Related
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
DBT	Maple syrup urine disease, type II
DHCR7	Smith-Lemli-Opitz syndrome
DLD	Dihydrolipoamide dehydrogenase deficiency
DMD	Duchenne muscular dystrophy
DYSF	Limb-girdle muscular dystrophy, type 2B; Miyoshi myopathy and distal myopathy with anterior tibial onset

GENE	CONDITION
EDA	Ectodermal dysplasia 1, hypohidrotic
ESCO2	Juberg-Hayward syndrome and Roberts-SC phocomelia syndrome
ETFA	Glutaric acidemia, Type IIA
ETFDH	Glutaric acidemia, Type IIC
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type I
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FKTN	Muscular dystrophy-dystroglycanopathy type 4A (Walker-Warburg syndrome); 4B, 4C; Cardiomyopathy, dilated, 1X
G6PC	Glycogen storage disease, type IA
GAA	Glycogen storage disease, type II
GALC	Krabbe disease
GALNS	Mucopolysaccharidosis IVA
GALT	Galactosemia
GBA	Gaucher disease
GCDH	Glutaric acidemia, type I
GJB2	Nonsyndromic hearing loss, GJB2-related
GJB6	Deafness, autosomal recessive 1B
GLA	Fabry disease
GLB1	GM1-gangliosidosis; Mucopolysaccharidosis type IVB
GLDC	Glycine encephalopathy, GLDC-related
GNPTAB	Mucolipidosis, type II/III alpha/beta;
GRHPR	Primary hyperoxaluria, type II
GUSB	Mucopolysaccharidosis VII
HADHA	Long-chain 3-Hydroxyacyl-CoA dehydrogenase deficiency; Trifunctional protein deficiency
HAX1	Severe congenital neutropenia 3, autosomal recessive
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, and other hemoglobinopathies
HEXA	Tay-Sachs disease; GM2-ganlgliosidosis
HEXB	Sandhoff disease
HGSNAT	Mucopolysaccharidosis, type IIIC
HMGCL	3-hydroxy-3-methylglutaryl CoA lyase deficiency
HYAL1	Mucopolysaccharidosis type IX
IDS	Mucopolysaccharidosis II
IDUA	Mucopolysachharidosis, type I (Hurler syndrome)
IVD	Isovaleric acidemia
MAN2B1	Abetalipoproteinemia
MCCC1	Usher syndrome, type 1B
MCCC2	Mucopolysaccharidosis, type IIIB (Sanfilippo B)
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis type IV
MEFV	Familial Mediterranean fever
MLC1	Megalencephalic Leukoencephalopathy with subcortical cysts, type I
MMAA	Methylmalonic aciduria, cblA type
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Methylmalonic aciduria, cblD type
MMUT	Methylmalonic aciduria, mut(0) type
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency

GENE	CONDITION
NBN	Nijmegen breakage syndrome
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NEB	Nemaline myopathy 2
NPC1	Niemann-pick disease, type C1
NPHS1	Nephrotic syndrome, type I
NPHS2	Nephrotic syndrome, type II
OXTR	Limb-Girdle Muscular Dystrophy Type 1C and Rippling Muscle Disease 2
PAH	Phenylalanine hydroxylase deficiency
PCCA	Propionic acidemia, PCCA-related
PCCB	Propionic acidemia, PCCB-related
PCDH15	Usher syndrome, type IF; Deafness, autosomal recessive 23
PEX1	Zellweger syndrome spectrum, PEX1-related
PEX7	Rhizomelic chondrodysplasia punctata, type I;
PKHD1	Polycystic kidney disease, autosomal recessive
PMM2	Congenital disorder of glycosylation, type IA
PPT1	Neuronal ceroid lipofuscinosis 1, PPT1-related
PRDX1	Methylmalonic aciduria and homocystinuria, cblC type
PRF1	Aplastic anemia; Hemophagocytic lymphohistiocytosis, familial, 2; Lymphoma, non-Hodgkin
PYGM	Glycogen storage disease, type V
RAG1	Omenn syndrome; Severe combined immunodeficiency
RPE65	Leber congenital amaurosis 2;
SACS	Spastic ataxia of Charlevoix-Saguenay, autosomal recessive
SAMHD1	Aicardi-Goutieres syndrome 5
SERPINA1	Alpha-1 antitrypsin deficiency
SGCA	Limb-girdle muscular dystrophy, type 2D
SGCB	Limb-girdle muscular dystrophy, type 2E
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A)
SLC22A5	Primary carnitine deficiency
SLC26A2	Sulfate transporter-related osteochondrodysplasia; Achondrogenesis Ib; Atelosteogenesis II; Diastrophic dysplasia; Epiphyseal dysplasia, multiple, 4
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct; Pendred syndrome
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease type A/B
STX11	Hemophagocytic lymphohistiocytosis, familial, 4
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5
TCIRG1	Osteopetrosis type 1, infantile malignant
TGM1	Lamellar ichthyosis type I
TH	Segawa syndrome (tyrosine hydroxylase deficiency)
TMEM216	Joubert syndrome 2; Meckel syndrome 2
TPP1	Neuronal ceroid lipofuscinosis 2, TPP1-related; Spinocerebellar ataxia type 7
TTPA	Ataxia with vitamin E deficiency
TYR	Albinism, oculocutaneous, type IA/IB; Waardenburg syndrome/albinism
UGT1A1	Hyperbilirubinemia, familial transient neonatal; Crigler-Najjar syndrome, type I/II
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3
USH2A	Usher syndrome, Type 2A; Retinitis pigmentosa 39
WAS	Neutropenia, severe congenital; Thrombocytopenia; Wiskott-Aldrich syndrome