Gene	Disease	Information	Residual risk in case of negative test result	Prevalence of carriers in the population
ABCA3	Surfactant metabolism dysfunction type 3	Surfactant metabolism dysfunction type 3 is a lung disorder caused by mutations in the ABCA3 gene. The ABCA3 gene provides instructions for making ABCA3 protein. The ABCA3 protein is found in the membrane that surrounds lamellar bodies, which are the cellular structures in which the phospholipids and proteins that make up surfactant are packaged. Without normal surfactant due to a mutations in the ABCA3 gene, alveoli sticks together after exhalation, causing them to collapse. The signs and symptoms of surfactant dysfunction are mostly problems with breathing and lack of oxygen. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:600 (Central Europe) and the risk of birth of an affected child is lower than 1:2400.	<1:600	1:30 (Central Europe)
ABCA4	Stargardt disease type 1	Stargardt disease type 1 is caused by mutations in the ABCA4 gene. The ABCA4 gene provides instructions for making the ABCA4 protein that transports potentially toxic substances out of photoreceptor cells. Mutations in the ABCA4 gene prevent the transportation and these toxic substances build up and form lipofuscin in the photoreceptor cells, eventually causing cell death. Loss of cells in the retina lead to progressive vision loss. Other symptoms of Stargardt disease are problems with night vision and impaired color vision. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1000 (Central Europe) and the risk of birth of an affected child is lower than 1:4000.	<1:1000	1:50 (Central Europe)
ABCC6	Pseudoxanthoma elasticum	Pseudoxanthoma elasticum (PXE) is a progressive disorder characterized by the accumulation of deposits of calcium and other minerals in elastic fibers. It is caused by mutations in the ABCC6 gene. This gene provides instructions for making a MRP6 protein. Mutations in the ABCC gene cause reduction of MRP6 protein, but is unclear how lack of properly functioning MRP6 protein leads to PXE. Signs and symptoms of PXE are papules in flexor areas, changes in the pigmented cells of the retina, bleeding and scarring the retina, or arteriosclerosis. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:112 (Central Europe) and the risk of birth of an affected child is lower than 1:448.	0,819444444	1:112 (Central Europe)
ABCC8	Neonatal diabetes mellitus Hyperinsulinemic hypoglycemia	The ABCC8 gene provides instructions for making the sulfonylurea receptor 1 (SUR1), which is one subunit of potassium channel that is found in the beta cells of the pancreas. This channel controls the secretion of insulin out of beta cells and into the bloodstream. Mutations in the ABCC8 gene can lead to neonatal diabetes mellitus (caused by reduced insulin secretion from beta cells) or to hyperinsulinemic hypoglycemia (caused by over-secretion of insulin from beta cells). In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2240 (Central Europe, 1:1040 for Ashkenazi) and the risk of birth of an affected child is lower than 1:8960 (1:4160 for Ashkenazi).	<1:2240 (Central Europe), <1:1040 (Ashkenazi)	1:112 (Central Europe) 1:52 (Ashkenazi)
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	Medium-chain acyl-CoA dehydrogenase deficiency is a condition caused by mutations in the ACADM gene. This gene provides instructions for making an enzyme which is required to metabolize a group of fats called medium-chain fatty acids. Mutations in the ACADM gene can cause deficiency of the enzyme and fatty acids are not converted into energy. This can lead to lack of energy and hypoglycemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1300 (Central Europe) and the risk of birth of an affected child is lower than 1:5200.	<1:1300	1:65 (Central Europe)

ACADS	Short-chain acyl-CoA dehydrogenase deficiency	Short-chain acyl-CoA dehydrogenase deficiency is a condition caused by mutations in the ACADS gene. This gene provides instructions for making an enzyme which is required to metabolize a group of fats called short-chain fatty acids. Mutations in the ACADS gene can cause deficiency of the enzyme and fatty acids are not converted into energy. This can lead to lack of energy and hypoglycemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2200 (Central Europe) and the risk of birth of an affected child is lower than 1:8800.	<1:2200	1:110 (Central Europe)
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	Very long-chain acyl-CoA dehydrogenase deficiency is a condition caused by mutations in the ACADVL gene. This gene provides instructions for making an enzyme which is required to metabolize a group of fats called very long-chain fatty acids. Mutations in the ACADVL gene can cause deficiency of the enzyme and fatty acids are not converted into energy. This can lead to lack of energy and hypoglycemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Central Europe)
AGL	Glycogen storage disease type III	Glycogen storage disease type III (also known as Cori disease) is an inherited disorder caused by mutations in the AGL gene. The AGL gene provides instructions for producing an enzyme called glycogen debranching enzyme, which helps break down glycogen. Mutations in the AGL gene prevent glycogen from being broken down effectively, which allows this sugar to build up to toxic levels in cells. Accumulation damages organs and tissues throughout the body, particularly the liver and muscles. Symptoms of this condition are enlarged liver, cirrhosis, adenomas in the liver, myopathy, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1400 (Central Europe) and the risk of birth of an affected child is lower than 1:5600.	<1:1400	1:70 (Central Europe)
AGXT	Primary hyperoxaluria type I	Primary hyperoxaluria type I is a rare condition caused by mutations in the AGXT gene. The AGXT gene provides instructions for making an enzyme called serine-pyruvate aminotransferase, which breaks down glyoxylate in peroxisomes. Mutations in the AGXT gene lead to convertion of glyoxylate into a substance called oxalate. Excess amounts of oxalate can lead to kidney and bladder stones and additional health problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2800 (Central Europe) and the risk of birth of an affected child is lower than 1:11200.	<1:2800	1:140 (Central Europe)
ALDOB	Hereditary fructose intolerance	Hereditary fructose intolerance is a condition caused by mutations in the ALDOB gene, normally producing enzyme aldolase B. ALDOB gene mutations reduce the function of the enzyme, impairing its ability to metabolize fructose. After ingesting fructose, individuals with hereditary fructose intolerance may experience nausea, bloating, abdominal pain, diarrhea, vomiting, and hypoglycemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1400 (Central Europe) and the risk of birth of an affected child is lower than 1:5600.	<1:1400	1:70 (Central Europe)
ALPL	Hypophosphatasia	Hypophosphatasia is an inherited disorder caused by mutations in the ALPL gene. This gene provides instructions for making an enzyme called tissue-nonspecific alkaline phosphatase, which plays an essential role in mineralization of the skeleton and teeth. A shortage of functional enzyme disrupts a mineralization and allows substances that are normally processed by the enzyme to build up in the body. The signs and symptoms of hypophosphatasia are skeletal abnormalities, early loss of baby teeth, hypercalcaemia, and kidney problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe) and the risk of birth of an affected child is lower than 1:12000.	<1:3000	1:270 (Central Europe) 1:150 (Canada)

AR	Androgen insensitivity syndrome	Androgen insensitivity syndrome is a condition that affects sexual development. Its is caused by mutations in the AR gene, which provides instructions for making an androgen receptor. As a result of mutations, cells that are sensitive to androgens become less responsive or unable to respond to these hormones at all. Even thought people with this condition are genetically male (with one X chromosome and one Y chromosome), they have mostly female sex characteristics, but do not have a uterus and therefore do not menstruate and are infertile. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:100000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:400000.	<1:100000	1:5000 (Central Europe) female carriers
ARSA	Metachromatic leukodystrophy	Metachromatic leukodystrophy is a genetic disorder caused by mutations in the ARSA gene. Mutations in the ARSA gene lead to the lack of an important enzyme called arylsulfatase A, which procces sulfatides in lysosomes. Because this enzyme is missing, sulfatides build up in the body and damage the nervous system, especially the cells that produce myelin. In people with metachromatic leukodystrophy, white matter damage causes progressive deterioration of intellectual functions and motor skills, seizures, paralysis, an inability to speak, blindness, and hearing loss. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Central Europe)
ASL	Argininosuccinic aciduria	Argininosuccinic aciduria is an inherited disorder caused by mutations in the ASL gene. The ASL gene provides instructions for making the enzyme argininosuccinate lyase, which participates in the urea cycle. If the argininosuccinate lyase enzyme is misshapen or missing due to a mutation in the ASL gene, it cannot fulfill its role in the urea cycle and excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. This buildup of ammonia damages the brain and other tissues and causes neurological problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2600 (Central Europe) and the risk of birth of an affected child is lower than 1:10400.	<1:2600	1:130 (Central Europe)
ASPA	Canavan disease	Canavan disease is a rare inherited disorder caused by mutations in the ASPA gene. The ASPA gene provides instructions for making an enzyme called aspartoacylase. This enzyme breaks down a compound called N-acetyl-L-aspartic acid (NAA), which is predominantly found in neurons in the brain. Mutations in the ASPA gene reduce the function of aspartoacylase, which allows NAA to build up to high levels in the brain. Affected infants usually do not develop motor skills and suffer from hypotonia and macrocephaly. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3160 (worldwide, 1:800 for Ashkenazi) and the risk of birth of an affected child is lower than 1:12640 (1:3200 for Ashkenazi).	<1:3160 (worldwide), <1:800 (Ashkenazi)	1:158 (worldwide),1:40 (Ashkenazi)
ASS1	Citrulinemia Type 1	Type I citrullinemia is an inherited disorder that is caused by mutations in the ASS1 gene. This gene provides instructions for making an enzyme argininosuccinate synthase 1, which is responsible for one step of the urea cycle. Mutations in the ASS1 gene reduce the activity of the enzyme, which disrupts the urea cycle and leads to accumulation of ammonia and other byproducts in the bloodstream. Ammonia is toxic to the nervous system. Symptoms of type I citrullinemia are lethargy, vomiting, seizures, and loss of consciousness. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2200 (Central Europe) and the risk of birth of an affected child is lower than 1:8800.	<1:2200	1:110 (Central Europe)

ATM	Ataxia-telangiectasia	Ataxia-telangiectasia is a rare inherited disorder caused by mutations in the ATM gene, which provides instructions for making a protein that helps control cell division and is involved in DNA repair. Without this protein, cells become unstable and die (especially cerebellum cells involved in coordinating movements). Mutations in the ATM gene also prevent cells from responding correctly to DNA damage, which allows breaks in DNA to accumulate. This can lead to the formation of cancerous tumors, particularly leukemia and lymphoma. This disorder is characterized by ataxia, chorea, and neuropathy. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Central Europe)
ATP7B	Wilson disease	Wilson disease is an inherited metabolic disorder caused by mutations in the ATP7B gene. This gene provides instructions for making a protein called copper-transporting ATPase 2, which plays a role in the transport of copper from the liver to other parts of the body. Mutations in the ATP7B gene prevent the transport protein from functioning properly and copper accumulates to toxic levels. Copper accumulation can lead to liver problems, neurological and psychiatric problems, and Kayser-Fleischer ring in cornea. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1800 (Central Europe) and the risk of birth of an affected child is lower than 1:8200.	<1:1800	1:90 (Central Europe)
BCKDHA	Maple syrup urine disease, type Ia	Maple syrup urine disease (type Ia) is an inherited disorder caused by mutations in the BCKDHA gene. The BCKDHA gene provides instructions for making the alpha subunit of the branched-chain alpha-keto acid dehydrogenase (BCKD) enzyme complex. Mutations in the gene reduce or eliminate the function of the enzyme complex, preventing the normal breakdown of leucine, isoleucine, and valine. As a result, toxic amounts of these amino acids and their byproducts build up in the body. Maple syrup urine disease gets its name from the distinctive sweet odor of affected infants' urine. It is also characterized by poor feeding, vomiting, lack of energy, abnormal movements, and delayed development. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:6400 and the risk of birth of an affected child is lower than 1:25600.	<1:6400	1:320 (Amish)
BCKDHB	Maple syrup urine disease, type Ib	Maple syrup urine disease (type Ib) is an inherited disorder caused by mutations in the BCKDHB gene. The BCKDHB gene provides instructions for making the beta subunit of the branched-chain alpha-keto acid dehydrogenase (BCKD) enzyme complex. Mutations in the gene reduce or eliminate the function of the protein complex, preventing the normal breakdown of leucine, isoleucine, and valine. As a result, toxic amounts of these amino acids and their byproducts build up in the body. Maple syrup urine disease gets its name from the distinctive sweet odor of affected infants' urine. It is also characterized by poor feeding, vomiting, lack of energy, abnormal movements, and delayed development. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:7200 and the risk of birth of an affected child is lower than 1:28800.	<1:7200	1:360 (Amish)
BCS1L	Mitochondrial complex III deficiency, GRACILE Syndrome, Björnstad Syndrome	The BCS1L gene provides instructions for making a protein that functions in mitochondria. The BCS1L protein is critical for the formation of a group of proteins known as complex III. This complex participates in oxidative phosphorylation, in which oxygen and simple sugars are used to create adenosine triphosphate (ATP), the cell's main energy source. Mutations in the BCS1L gene are related to several health conditions: Mitochondrial complex III deficiency, GRACILE syndrome, and Björnstad syndrome. Symptoms of these syndroms are linked to abnormal function of mitochondria. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2240 (Central Europe) and the risk of birth of an affected child is lower than 1:8960.	<1:2240	1:112 (Central Europe)

BLM	Bloom syndrome	Bloom syndrome is an inherited disorder caused by mutations in the BLM gene. The BLM gene provides instructions for making a member of a protein family called RecQ helicases. The BLM protein helps to prevent excess sister chromatid exchanges and it also helps maintain the stability of the DNA during the replication. BLM gene mutations lead to the absence of functional BLM protein and as a result, the sister chromatid exchange and chromosome breakage occurs more frequently. The cells are also less able to repair DNA damage caused by ultraviolet ligh. Syptoms of Bloom syndrome are reddened skin, skin rashes hypo- or hyperpigmentation and increased risk of cancer. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1571 (Ashkenazi) and the risk of birth of an affected child is lower than 1:6284.	<1:1571	1:110 (Ashkenazi)
BTD	Biotinidase deficiency	Biotinidase deficiency is an inherited disorder caused by mutations in the BTD gene. The BTD gene provides instructions for making an enzyme called biotinidase. This enzyme recycles biotin and mutations in the BTD gene reduce or eliminate its function. Biotin is needed to break down fats, proteins, and carbohydrates and the shortage of free biotin leads to a buildup of potentially toxic compounds in the body. Biotinidase deficiency can cause seizures, hypotonia, breathing problems, hearing and vision loss, ataxia, skin rashes, alopecia, and candidiasis. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2400 (Central Europe) and the risk of birth of an affected child is lower than 1:9600.	<1:2400	1:120 (Central Europe)
CAPN3	Limb-girdle muscular dystrophy type 2A	Limb-girdle muscular dystrophy type 2A, also known as calpainopathy, is caused by mutations in the CAPN3 gene. The CAPN3 gene provides instructions for making an enzyme called calpain-3, which is found within muscle cells in sarcomeres. Mutations in the gene disrupt muscle maintenance and repair. The muscles most affected are muscles of the shoulders, upper arms, pelvic area, and thighs. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2400 (Central Europe, 1:2000 for Nothern Italy) and the risk of birth of an affected child is lower than 1:9600 (1:8000 for Nothern Italy).	<1:2000 (Northern Italy), <1:2400 (Central Europe)	1:100 (Northern Italy), 1:120 (Central Europe)
CBS	Homocystinuria	Homocystinuria is an inherited metabolic disorder caused by mutations in CBS gene. The CBS gene provides instructions for making an enzyme called cystathionine beta-synthase, which converts homocysteine and serine to a molecule called cystathionine. Mutations in CBS gene disrupt normal function of this enzyme and as a result, toxic level of homocysteine builds up in the blood and urine. Homocystinuria has several features in common with Marfan syndrome, including skeletal and eye changes. Other symptoms are related to connective tissue, muscles, central nervous system, and cardiovascular system. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1400 (Central Europe) and the risk of birth of an affected child is lower than 1:5600.	<1:1400	1:70 (Central Europe)
CFTR	Cystic fibrosis	Cystic fibrosis is a disorder caused by mutations in the CFTR gene. Mutations in the CFTR gene alter the production, structure, or stability of the chloride channel. All of these changes prevent the channel from functioning properly, which impairs the transport of chloride ions and the movement of water into and out of cells. As a result, cells that line the passageways of the lungs, pancreas, and other organs, produce mucus that is abnormally thick and sticky. The abnormal mucus obstructs the airways and glands. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:750 (Central Europe) and the risk of birth of an affected child is lower than 1:3000.	<1:750	1:25 (Central Europe)

CLN3 (1 breakpoint)	Neuronal ceroid lipofuscinosis type 3	Neuronal ceroid lipofuscinosis type 3 is an inherited disorder caused by mutations in the CLN3 gene, which encodes CLN3 protein. Exact function of this protein and how mutations lead to the disease is unclear. This disorder is characterized by accumulation of proteins and other substances in lysosomes, which can cause death of nerve cells. Neurological signs and symptoms are as vision impairment, intellectual disability, movement problems, speech difficulties and seizures. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:6000 (Central Europe) and the risk of birth of an affected child is lower than 1:24000.	<1:6000	1:180 (Central Europe)
CNGB3	Achromatopsia	Achromatopsia is a disorder of the retina and it is caused by mutations in CNGB3 gene. The CNGB3 gene provides instructions for making one part (the beta subunit) of the cone ion channel. These channels are found exclusively in cones, which are located in retina. Cones provide vision in bright light and color vision. Mutations in the CNGB3 gene prevent cones from reacting appropriately to light. This condition can lead topartial or total absence of color vision. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2240 (Central Europe) and the risk of birth of an affected child is lower than 1:8960.	<1:2240	1:112 (Central Europe)
COL4A3	Alport syndrome 2	Alport syndrome 2 is a genetic condition caused by mutations in the COL4A3 gene. The COL4A3 makes the alpha3(IV) chain of type IV collagen. This protein plays an important role in the kidneys, inner ear structures and the eye. Mutations in the COL4A3 gene result in serious deficiency of the type IV collagen. People with Alport syndrome experience progressive loss of kidney function and often have blood and proteins in their urine. They also frequently develop hearing and vision loss. These symptoms are more common in males with Alport syndrome than in affected females. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:5400 (Central Europe) and the risk of birth of an affected child is lower than 1:21600.	<1:5400	1:270 (Central Europe)
COL4A5	Alport syndrome 1	Alport syndrome is a genetic condition caused by mutations in the COL4A5 gene. The COL4A5 gene makes the alpha5(IV) chain of type IV collagen. This protein plays an important role in the kidneys, inner ear structures and the eye. Mutations in the COL4A5 gene result in serious deficiency of the type IV collagen. People with Alport syndrome experience progressive loss of kidney function and often have blood and proteins in their urine. They also frequently develop hearing and vision loss. These symptoms are more common in males with Alport syndrome than in affected females. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:300000 (worldwide, for women) and the risk of birth of an affected child is lower than 1:1200000.	<1:300000	1:30000 females (Central Europe)
COL7A1	Dystrophic epidermolysis bullosa	Dystrophic epidermolysis bullosa is a genetic condition caused by mutations in the COL7A1 gene. The COL7A1 makes the alpha1(VII) chain of type VII collagen. This protein plays an important role in strengthening and stabilizing the skin. Mutations in the COL7A1 gene result in serious deficiency of the type VII collagen, which impairs the connection between the epidermis and the dermis and causes the extreme skin fragility. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:530 (Czechia) and the risk of birth of an affected child is lower than 1:2120.	<1:530	1:370 (Czech Republic)

CPT2	Carnitine palmitoyltransferase II deficiency	Carnitine palmitoyltransferase II deficiency is a condition that prevents the body from using certain fats for energy and is caused by mutations in the CPT2 gene. This gene provides instructions for making an enzyme called carnitine palmitoyltransferase 2, which is essential for fatty acid oxidation. Mutations in the CPT2 gene disrupt the function of enzyme and cause muscle pain and weakness, arhythmia, hypoglycemia and hypoketosis. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (worldwide, 1:800 for Ashkenazi) and the risk of birth of an affected child is lower than 1:12800 (1:3200 for Ashkenazi).	<1:800 (Ashkenazi), <1:3200 (worldwide)	1:40 (Ashkenazi), 1:160 (worldwide)
CTNS	Cystinosis	Cystinosis is a condition characterized by accumulation of the amino acid cystine within cells and it is caused by mutations in CTNS gene. This gene provides instructions for making a transporter protein called cystinosin, which normally moves cystine out of the lysosomes. Mutations in the CTNS gene disrupt this function and excess cystine accumulates and forms crystals in the lysosome. The buildup of cystine damages cells in the kidneys and eyes and may also affect other organs. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe, 1:1600 for Bretagne) and the risk of birth of an affected child is lower than 1:12000 (1:6400 for Bretagne).	<1:3000, <1:1600	1:150 (Central Europe) 1:80 (Brittany)
CYP21A2	Congenital adrenal hyperplasia (21-hydroxylase deficiency)	Congenital adrenal hyperplasia (21-hydroxylase deficiency) is an inherited disorder that caused by mutations in the CYP21A2 gene. The CYP21A2 gene provides instructions for making an enzyme called 21-hydroxylase. This enzyme plays role in producing cortisol and aldosteron. With 21-hydroxylase deficiency, substances that are usually used to form these hormones build up in the adrenal glands and are converted to androgens instead. Excess production of androgens leads to abnormalities of sexual development. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:180 (Central Europe) and the risk of birth of an affected child is lower than 1:720.	<1:180	1:45 (Central Europe)
CYP27A1	Cerebrotendinous xanthomatosis	Cerebrotendinous xanthomatosis is a neurological disorder caused by mutations in the CYP27A1 gene, which produces an enzyme called sterol 27-hydroxylase. This enzyme is located in mitochondria and involved in break down of cholesterol. People with this disorder cannot break down cholesterol effectively, so it accumulate in the body in the form of xanthomas. These xanthomas are most commonly found in the brain and in tendon, where they cause various neurological problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2200 (Central Europe) and the risk of birth of an affected child is lower than 1:8800.	<1:2200	1:110 (Central Europe)
DHCR7	Smith-Lemli-Opitz syndrome	Smith-Lemli-Opitz syndrome is a developmental disorder caused by mutations in the DHCR7 gene. The DHCR7 gene provides instructions for making an enzyme called 7-dehydrocholesterol reductase, which is responsible for the final step in cholesterol production. Mutations in the DHCR7 gene reduce or eliminate the activity of this enzyme, preventing cells from producing enough cholesterol and allowing toxic byproducts of cholesterol production to build up in the body. This combination disrupts the growth and development of many body systems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1000 (Central Europe) and the risk of birth of an affected child is lower than 1:4000.	<1:1000	1:50 (Central Europe)

DLD	Dihydrolipoamide dehydrogenase deficiency	Dihydrolipoamide dehydrogenase deficiency is a severe condition caused by mutations in the DLD gene. DLD gene encodes part of an enzyme complex that breaks down leucine, isoleucin and valin. Mutations in the DLD gene impair the function of the DLD enzyme. As a result, molecules that are normally broken down and their byproducts build up in the body, damaging tissues and leading to lactic acidosis and other chemical imbalances. Especially affected are brain and liver. Neurological problems are also common in this condition. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Ashkenazi) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Ashkenazi)
DMD	Duchenne and Becker muscular dystrophy	Duchenne and Becker muscular dystrophies have similar signs and symptoms and are caused by different mutations in the same gene. DMD provides instructions for making a protein called dystrophin, which is located primarily in skeletal and cardiac muscles. Both the Duchenne and Becker forms of muscular dystrophy are associated with muscle weakness and wasting and cardiomyopathy. The two conditions differ in their severity, age of onset, and rate of progression and they occur almost exclusively in males. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:30000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:120000.	<1:30000	1:3000 females (Central Europe)
ELP1	Familial dysautonomia	Familial dysautonomia is a genetic disorder caused by mutations in the ELP1 gene. The ELP1 gene encodes a protein that is found in brain cells and is important for the transcription of proteins that affect the cytoskeleton and cell movement. Cytoskeleton plays a critical role in the growth of nerve cells and cell motility is crucial for the movement of nerve cells to their proper locations in the brain. Mutations in the ELP1 gene disrupt all of these functions. The disorder disturbs cells in the autonomic nervous system, which controls digestion, breathing, and the regulation of blood pressure and body temperature. It also affects the sensory nervous system, which controls taste and the perception of pain, heat, and cold. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:600 (Ashkenazi) and the risk of birth of an affected child is lower than 1:2400.	<1:600 (Ashkenazi)	1:30 (Ashkenazi)
F11	Factor XI deficiency	Factor XI deficiency is a bleeding disorder caused by mutations in the F11 gene. The F11 gene provides instructions for making a protein called factor XI. Mutations in the F11 gene lead to the production of none or an abnormal version of factor XI, which can no longer participate effectively in the blood clotting process. The most common feature of factor XI deficiency is prolonged bleeding after trauma or surgery, especially involving the inside of the mouth and nose or the urinary tract. Other signs and symptoms can include frequent nosebleeds, easy bruising, bleeding under the skin, and bleeding of the gums. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:750 (Ashkenazi) and the risk of birth of an affected child is lower than 1:3000.	<1:750 (Ashkenazi)	up to 1:15 (Ashkenazi)
F8	Hemophilia A	Hemophilia A is a bleeding disorder caused by changes in the F8 gene. The F8 gene provides instructions for making a protein called coagulation factor VIII. Mutations in F8 gene lead to the production of none or an abnormal version of coagulation factor VIII, which can no longer participate effectively in the blood clotting process. Problems with blood clotting lead to continuous bleeding that can be difficult to control. Common signs are spontaneous bleeding into the urine, gastrointestinal tract, skull cavity, or bleeding into the muscles and joints. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:10000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:40000.	<1:10000	1:5000 females (Central Europe)

F9	Hemophilia B	Hemophilia B is a bleeding disorder caused by changes in the F9 gene. The F9 gene provides instructions for making a protein called coagulation factor IX. Mutations in F8 gene lead to the production of none or an abnormal version of coagulation factor IX, which can no longer participate effectively in the blood clotting process. Problems with blood clotting lead to continuous bleeding that can be difficult to control. Common signs are spontaneous bleeding into the urine, gastrointestinal tract, skull cavity, or bleeding into the muscles and joints. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:400000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:1600000.	<1:400000	1:20000 females (Central Europe)
FAH	Tyrosinemia type I	Tyrosinemia type I is a metabolic disorder caused by mutations in the FAH gene. The FAH gene provides instructions for making an enzyme called fumarylacetoacetate hydrolase, which breaks down tyrosine. Mutations in the FAH gene cause a decrease in the enzyme activity. As a result, tyrosine and its byproducts accumulate to toxic levels, which can cause damage and death to cells in the liver, kidneys, nervous system, and other organs. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe) and the risk of birth of an affected child is lower than 1:12000.	<1:3000	1:150 (Central Europe)
FANCA	Fanconi anemia type A	Fanconi anemia type A is a condition caused by mutations in one of three major genes (FANCA, FANCC, FANCG). Proteins produced from these genes are involved in a cell process known as the FA pathway, which participates in interstrand cross-links (ILCs) repair. Mutations in any of these genes will disrupt the entire FA pathway. As a result, DNA damage is not repaired efficiently and ICLs build up over time. People with this condition may have bone marrow failure, physical abnormalities, organ defects, and an increased risk of certain cancers. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2500 (worldwide) and the risk of birth of an affected child is lower than 1:10000.	<1:2500	1:250 (worldwide)
FANCC	Fanconi anemia type C	Fanconi anemia is a condition caused by mutations in one of three major genes (FANCA, FANCC, FANCG). Proteins produced from these genes are involved in a cell process known as the FA pathway, which participates in interstrand cross-links (ILCs) repair. Mutations in any of these genes will disrupt the entire FA pathway. As a result, DNA damage is not repaired efficiently and ICLs build up over time. People with this condition may have bone marrow failure, physical abnormalities, organ defects, and an increased risk of certain cancers. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:400 (worldwide) and the risk of birth of an affected child is lower than 1:1600.	<1:400	1:200 (worldwide)
FMR1	Fragile X syndrome	Fragile X syndrome is a genetic condition caused by the mutations in the FMR1 gene, which encodes a protein called FMRP. This protein plays a role in the development of synapses, which are specialized connections between nerve cells. Synapses are critical for relaying nerve impulses. Expansion of CGG triplet silences the FMR1 gene, which prevents the gene from producing FMRP. Deficiency of this protein disrupts nervous system functions and leads to the signs and symptoms of fragile X syndrome, such as intellectual disability, anxiety and hyperactive behavior, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:80000 (worldwide, for women) and the risk of birth of an affected child is lower than 1:320000.	<1:80000	1:4000 females (worldwide)

G6PC	Glycogen storage disease type 1a	Glycogen storage disease type I (also known as von Gierke disease) is an inherited disorder caused by mutations in the G6PC gene. Proteins produced from the G6PC gene break down a glucose 6-phosphate into glucose. Due to a mutations in the G6PC gene, glucose 6-phosphate is not broken down into glucose, but converted to glycogen and fat so it can be stored within cells. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3160 (worldwide) and the risk of birth of an affected child is lower than 1:12640.	<1:3160	1:158 (worldwide)
GAA	Glycogen storage disease II (Pompe disease)	Glycogen storage disease II (also known as Pompe disease) is an inherited disorder caused by mutations in the GAA gene. The GAA gene provides instructions for producing an enzyme called acid alpha-glucosidase, which breaks down glycogen into glucose. Mutations in the GAA gene prevent acid alpha-glucosidase from breaking down glycogen effectively, which allows this sugar to build up to toxic levels in lysosomes. This buildup damages organs and tissues throughout the body, particularly the muscles. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2400 (Central Europe) and the risk of birth of an affected child is lower than 1:9600.	<1:2400	1:120 (Central Europe)
GALC	Krabbe disease	Krabbe disease is a severe neurological condition caused by mutations in the GALC gene. Enzyme galactosylceramidase produced by GALC gene breaks down galactolipids. Some of these lipids are important components of myelin. Because of mutations in the GALC gene and reduced activity of galactosylceramidase, excess galactolipids are accumulated in certain cells, which can cause damage to myelin-forming cells, eventually leading to demyelination in the nervous system. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe) and the risk of birth of an affected child is lower than 1:12000.	<1:3000	1:150 (Central Europe)
GALT	Galactosemia	Galactosemia is a metabolic condition caused by mutations in the GALT gene. GALT gene produces enzyme galactose-1-phosphate uridylyltransferase, which enables the body to process a galactose. Due to a mutations in the GALT gene and reduced activity of the enzyme, excess galactose and other byproducts build up to toxic levels in the body. The accumulation of these substances damages tissues and organs. Symptoms of galactosemia are feeding difficulties, lethargy, yellowing of the skin and whites of the eyes, liver damage, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Central Europe)
GBA	Gaucher disease, type I	Gaucher disease is an inherited disorder caused by mutations in the GBA gene. The GBA gene provides instructions for making an enzyme called beta-glucocerebrosidase, which breaks down glucocerebroside into a glucose and a ceramide. Due to a mutations in the GBA gene and reduced activity of the enzyme, excess glucocerebroside and related substances can build up to toxic levels within cells, damaging tissues and organs. Symptoms of Gaucher disease are enlargement of the liver and spleen, anemia, easy bruising, lung disease, and bone abnormalities. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1100 (Central Europe) and the risk of birth of an affected child is lower than 1:4400.	<1:1100	1:110 (Central Europe)
GCDH	Glutaric aciduria, type I	Glutaric aciduria type I is an inherited disorder caused by mutations in the GCDH gene. The GCDH gene provides instructions for making the enzyme glutaryl-CoA dehydrogenase. This enzyme is involved in processing the amino acids lysine, hydroxylysine and tryptophan. A shortage of this enzyme allows these amino acids and their intermediate breakdown products to build up to abnormal levels, which can damage the brain, particularly the basal ganglia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1800 (Central Europe) and the risk of birth of an affected child is lower than 1:7200.	<1:1800	1:90 (Central Europe)

GJB2	Deafness, autosomal recessive 1A	Nonsyndromic hearing loss, inherited in an autosomal recessive pattern, is a partial or total loss of hearing caused by mutations in the GJB2 gene. Mutations in the GJB2 gene alter a protein called connexin 26, which changes the structure of gap junctions and may affect the function or survival of cells that are needed for hearing. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:670 (Central Europe) and the risk of birth of an affected child is lower than 1:2680.	<1:670	1:20 (Central Europe)
GLA	Fabry disease	Fabry disease is an inherited metabolic disorder caused by mutations in the GLA gene. This gene provides instructions for making an enzyme called alpha-galactosidase A. Mutations in the GLA gene alter the structure and function of this enzyme, which leads to accumulation of globotriaosylceramide in cells throughout the body. Particularly in cells lining blood vessels in the skin and cells in the kidneys, heart, and nervous system. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1000000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:4000000.	<1:1000000	1:50000 females (Central Europe)
GLB1	GM1 gangliosidosis	GM1 gangliosidosis is an inherited disorder caused by mutations in the GLB1 gene. The GLB1 gene provides instructions for making an enzyme called beta-galactosidase, which breaks down GM1 ganglioside. Mutations in the GLB1 gene reduce or eliminate the activity of β-galactosidase and GM1 ganglioside can not be broken down. As a result, this substance accumulates to toxic levels in many tissues and organs, particularly in the brain, leading to the destruction of nerve cells in the brain. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe, 1:1000 for Roma populations) and the risk of birth of an affected child is lower than 1:12800 (1:4000 for Roma populations).	<1:3200, <1:1000	1:160 (Central Europe), 1:50 (Romani people)
GNPTAB	Mucolipidosis type II and III	Mucolipidosis type II and III are disorders caused by mutations in the GNPTAB gene. This gene provides instructions for making a part of an enzyme called GlcNAc-1-phosphotransferase. GlcNAc-1-phosphotransferase is involved in the process of attaching a mannose-6-phosphate to specific digestive enzymes. Mutations in the GNPTAB gene disrupt the tagging, which prevents many enzymes from reaching the lysosomes. The signs and symptoms of mucolipidosis II and III are most likely due to the shortage of digestive enzymes inside lysosomes. Some of them are bone abnormalities, heart valve abnormalities, hoarse voice, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe, 1:600 for Quebec) and the risk of birth of an affected child is lower than 1:12800 (1:2400 for Quebec).	<1:3200, <1:600	1:160 (Central Europe) 1:30 (Quebec)
HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase deficiency	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency is a condition caused by mutations in the HADHA gene. This gene provides instructions for making one part of an enzyme complex, which is required for fatty acid oxidation in mitochondria. Mutations in the HADHA gene can cause deficiency of the enzyme complex and long-chain fatty acids are not converted into energy. This can lead to lack of energy and hypoglycemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:850 (Central Europe) and the risk of birth of an affected child is lower than 1:3400.	<1:850	1:110 (Central Europe)

HBA1/HBA2	Alpha-thalassemia	Alpha thalassemia is a disorder caused by deletions involving the HBA1 and HBA2 genes. Both of these genes provide instructions for making a protein called alpha-globin, which is a component of hemoglobin. A shortage of alpha-globin prevents cells from making normal hemoglobin. Instead, cells produce abnormal forms of hemoglobin called hemoglobin Bart (Hb Bart) or hemoglobin H (HbH). These abnormal molecules cannot effectively carry oxygen to the body's tissues, which causes an anemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:600 (Afro-Americans, 1:800 for Mediterranean, 1:400 for SE Asia) and the risk of birth of an affected child is lower than 1:2400 (1:3600 for Mediterranean, 1:1600 for SE Asia).	<1:600 (Afroamerican), <1:800 (Mediterranean), <1:400 (Southeast Asia)	1:30 (Afroamerican), 1:40 (Mediterranean), 1:20 (Southeast Asia)
HBB	Sickle cell anemia, Beta thalassemia	The HBB gene provides instructions for making a protein called beta-globin, a component of a hemoglobin. Due to a mutations in the HBB gene, an abnormal form of hemoglobin or a reduced amount of functional hemoglobin is produced, which disrupts the normal development of red blood cells and cause anemia. Disease caused by an abnormal version of beta-globin known as hemoglobin S (HbS) is called Sickle cell anemia. Disease caused by reduced amount of hemoglobin is called Beta-thalassemia (β0 thalassemia if none hemoglobin is produced, β+ thalassemia if a reduced amount is produced). In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1000 (Africans, Sickle cell anemia; 1:2000 Europe, Beta thalassemia) and the risk of birth of an affected child is lower than 1:4000 (Africans, Sickle cell anemia; 1:8000 Europe, Beta thalassemia).	<1:1000 (African or Afroamerican origin, sicklecell disease); <1:2000 (beta thalassaemia)	up to 1:10 (Central Africa, sickle cell anemia); 1:100 (Europe, beta thalasemia)
HEXA	Tay-Sachs disease	Tay-Sachs disease is a rare inherited disorder caused by mutations in the HEXA gene. The HEXA gene encodes a part of an enzyme called beta-hexosaminidase A, which helps break down GM2 ganglioside. Mutations in the HEXA gene disrupt the activity of this enzyme, which prevents the GM2 ganglioside from being broken down. As a result, this substance accumulates to toxic levels, particularly in neurons in the brain and spinal cord. Progressive destruction of the neurons lead to seizures, vision and hearing loss, intellectual disability, and paralysis. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:5400 (Central Europe, 1:600 for Ashkenazi) and the risk of birth of an affected child is lower than 1:21600 (1:4000 for Ashkenazi).	<1:600 (Askhenazi), <1:5400 (Central Europe)	1:30 (Ashkenazi), 1:270 (Central Europe)
IDUA	Mucopolysaccharidosis type I	Mucopolysaccharidosis type I is a condition caused by mutations in the IDUA gene. The IDUA gene provides instructions for producing an alpha-L-iduronidase enzyme that is involved in the breakdown of glycosaminoglycans (GAGs). Mutations in the IDUA gene reduce or completely eliminate the function of the IDUA enzyme, which leads to the accumulation of GAGs within cells, specifically inside the lysosomes. The accumulation of GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in this disorder. Other symptoms are macrocephaly, hydrocephalus, heart valve abnormalities or a large tongue. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3160 (Central Europe) and the risk of birth of an affected child is lower than 1:12640.	<1:3160	1:25000
IL2RG	X-linked severe combined immunodeficiency	X-linked severe combined immunodeficiency is an inherited disorder of the immune system caused by mutations in the IL2RG gene. The IL2RG gene provides instructions for making a protein that is necessary for the growth and maturation of developing lymphocytes. Mutations in the IL2RG gene prevent these cells from developing and functioning normally. Without functional lymphocytes, the body is unable to fight off infections. Other symptoms are chronic diarrhea, a fungal infection called thrush, and skin rashes. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:250000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:1000000.	<1:250000	1:25000 females (Central Europe)

LAMB3	Epidermolysis bullosa, junctional	Junctional epidermolysis bullosa is an inherited disorder caused by mutations in the LAMB3 gene. The LAMB3 gene provides instructions for making one part of a protein called laminin 332, which plays an important role in strengthening and stabilizing the skin by helping to attach the epidermis to underlying layers. Mutations in the LAMB3 gene lead to the production of a defective or nonfunctional version of this protein. Without functional laminin 332, the skin fragile, easily damaged and blisters form easily. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:5000 (Central Europe) and the risk of birth of an affected child is lower than 1:20000.	<1:5000	1:250 (Central Europe)
MCCC1	3-methylcrotonyl-CoA carboxylase deficiency 1	3-methylcrotonyl-CoA carboxylase deficiency I is an inherited disorder caused by mutations in the MCCC1 gene. This gene provide instructions for making one subunit of an enzyme called 3-methylcrotonyl-coenzyme A carboxylase (3-MCC), which is responsible for processing leucine. Mutations in the MCCC1 gene reduce or eliminate the activity of 3-MCC, preventing the body from processing leucine properly. As a result, toxic byproducts of leucine processing build up to harmful levels, which can damage the brain. The characteristic features of this condition include recurrent episodes of vomiting and diarrhea, lethargy, and hypotonia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1600 (Central Europe) and the risk of birth of an affected child is lower than 1:6400.	<1:1600	1:80 (Central Europe)
MCCC2	3-methylcrotonyl-CoA carboxylase deficiency 2	3-methylcrotonyl-CoA carboxylase deficiency II is an inherited disorder caused by mutations in the MCCC2 gene. This gene provide instructions for making one subunit of an enzyme called 3-methylcrotonyl-coenzyme A carboxylase (3-MCC), which is responsible for processing leucine. Mutations in the MCCC2 gene reduce or eliminate the activity of 3-MCC, preventing the body from processing leucine properly. As a result, toxic byproducts of leucine processing build up to harmful levels, which can damage the brain. The characteristic features of this condition include recurrent episodes of vomiting and diarrhea, lethargy, and hypotonia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1600 (Central Europe) and the risk of birth of an affected child is lower than 1:6400.	<1:1600	1:80 (Central Europe)
MCOLN1	Mucolipidosis type IV	Mucopolysaccharidosis type I is a condition caused by mutations in the MCOLN1 gene. The MCOLN1 gene provides instructions for producing a mucolipin-1 enzyme that transports lipids and proteins between lysosomes and endosomes. Mutations in the MCOLN1 gene result in the production of none or a nonfunctional protein, which impairs the transportation, causing these substances to build up inside lysosomes. Signs and symptoms of this condition are delayed development of mental and motor skills, problems with vision, achlorhydria, and anemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe, higher in Ashkenazi) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:100 (Ashkenazi)
MEFV	Familial Mediterranean fever	Familial Mediterranean fever is an inherited condition caused by mutations in the MEFV gene. The MEFV gene provides instructions for making a protein called pyrin, which likely assists in keeping the inflammation process under control. Mutations in the MEFV gene disrupts control of the inflammation process. Signs and symptoms of familial mediterranean fever are recurrent episodes of painful inflammation in the abdomen, chest, or joints, often accompanied by fever and sometimes a rash or headache. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:540 (Central Europe) and the risk of birth of an affected child is lower than 1:2160.	<1:540	1:16 (Europe)

MMACHC	Methylmalonic acidemia with homocystinuria cb1C type	Methylmalonic acidemia with homocystinuria cb1C type s an inherited disorder caused by mutations in the MMACHC gene. The MMACHC gene provides instructions for making a protein that helps convert vitamin B12 into adenosylcobalamin (AdoCbl). AdoCbl is required for the normal function of an enzyme that helps break down certain amino acids, lipids, and cholesterol. Mutations in the MMACHC gene affect early steps of vitamin B12 processing, resulting in a shortage of both AdoCbl. Without AdoCbl, proteins and lipids are not broken down properly and are accumulated in the body's organs and tissues instead. Signs and symptoms are failure to thrive, pale appearance, hypotonia, seizures, microcephaly, and dealyed development. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2800 (worldwide) and the risk of birth of an affected child is lower than 1:11200.	<1:2800	1:140 (worldwide)
MTM1	X-vázaná myotubulární myopatie	X-linked myotubular myopathy is a condition caused by mutations in the MTM1 gene. The MTM1 gene provides instructions for producing an enzyme called myotubularin, which is thought to be involved in the development and maintenance of muscle cells. MTM1 gene mutations disrupt function of myotubularin, causing muscle weakness and other signs and symptoms of X-linked myotubular myopathy, such as hypotonia, impaired development of motor skills, breathing problems, fragile bones, scoliosis and joint deformities of the hips and knees. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:250000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:1000000.	<1:250000	1:25000 females (Central Europe)
MUT	Methylmalonic aciduria mut(0) type	Methylmalonic aciduria mut(0) type is an inherited disorder caused by mutations in the MUT gene. This gene provides instructions for making an enzyme called methylmalonyl CoA mutase, which together with vitamin B12 breaks down amino acids, certain lipids, and cholesterol. Mutations in the MUT gene alter the enzyme's structure or reduce its amount, preventing these molecules from being broken down properly. Instead, they can accumulate in the body's organs and tissues. Signs and symptoms of Methylmalonic acidemia are delayed development, lethargy, hepatomegaly, kidney disease, and inflammation of the pancreas. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3020 (worldwide) and the risk of birth of an affected child is lower than 1:12080.	<1:3020	1:151 (worldwide)
MVK	Mevalonic aciduria	Mevalonic aciduria is an disorder caused by mutations in the MVK gene. Mutations in the MVK gene lead to deficiency of mevalonate kinase, which disrupt the biosynthesis of cholesterol and isoprenoids. Mevalonic acid, a precursor of cholesterol, is build up in the cells instead of being used to synthesize cholesterol and isoprenoids. Mevalonic aciduria is characterized by developmental delay, ataxia, epilepsy, progressive problems with vision, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:4480 (worldwide) and the risk of birth of an affected child is lower than 1:17920.	<1:4480	1:224 (worldwide), 1:65 Dutch
MYO7A	Usher syndrome type Ib	Usher syndrome type Ib is a condition caused by mutations in the MYO7A gene. The MYO7A gene provide instructions for making myosin VIIA protein, which is made in the inner ear and in the retina. Most of the gene mutations responsible for Usher syndrome lead to a loss of hair cells in the inner ear and a gradual loss of rods and cones in the retina. Degeneration of these sensory cells causes the hearing loss, balance problems, and vision loss. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2800 (Europe) and the risk of birth of an affected child is lower than 1:11200.	<1:2800	1:140 (Europe)

NAGLU	Mucopolysaccharidosis type IIIB	Mucopolysaccharidosis type IIIB is a progressive disorder caused by mutations in the NAGLU gene. The NAGLU gene provides instructions for producing an alpha-N-acetylglucosaminidase enzyme that is involved in the breakdown of heparan sulfate. Mutations in the NAGLU gene reduce or eliminate the function of the enzyme and the breakdown of a heparan sulfate is disrupted. As a result, partially broken down heparan sulfate accumulates within lysosomes, which negatively affects the brain and spinal cord, causing neurodegeneration. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2640 (worldwide) and the risk of birth of an affected child is lower than 1:10560.	<1:2640	1:132 (worldwide)
NBN	Nijmegen breakage syndrome	Nijmegen breakage syndrome is a condition caused by mutations in the NBN gene. The NBN gene provides instructions for making a protein called nibrin. This protein is involved in the repair of damaged DNA and regulation of cell division and proliferation. The NBN gene mutations lead to the production of an abnormally short version of the nibrin protein, which prevents it from responding to DNA damage effectively. As a result, affected individuals are highly sensitive to the radiation and other agents that can cause breaks in DNA. A buildup of these breaks increases the risk of cancer, specially of non-Hodgkin lymphoma. Other symptoms are distinctive facial features, and immunodeficiency. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Slavic population) and the risk of birth of an affected child is lower than 1:12000.	<1:3000 (Slavic population)	1:150 (Slavic population)
NPC1	Niemann-Pick disease type C1, D	Niemann-Pick disease type C1 and D is a condition caused by mutations in the NPC1 gene. The NPC1 gene provides instructions for making a protein that is involved in the movement of lipids within cells. Mutations in NPC1 gene lead to a shortage of functional protein, which prevents movement of cholesterol and other lipids, leading to their accumulation in cells and damage of tissues and organs. The signs and symptoms are ataxia, dystonia, inability to move the eyes vertically, problems with speech and swalowing, liver disease, and interstitial lung disease. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:4600 (Europe) and the risk of birth of an affected child is lower than 1:18400.	<1:4600	1:230 (Europe)
NPHS2	Congenital nephrotic syndrome type 2	Congenital nephrotic syndrome is a kidney condition caused by mutations in the NPHS2 gene. This gene provide instructions for making protein called podocin, which is found in cells called podocytes, located in glomeruli. Without a functional podocin, the filtering ability of the kidneys is impaired, causing more molecules get excreted in urine. Signs and symptoms of this condition are proteinuria, hypercholesterolemia, buildup of fluid and swelling in the abdominal cavity, and anemia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe, 1:1000 for Finland) and the risk of birth of an affected child is lower than 1:12800 (1:4000 for Finland).	<1:1000 (Finland), <1:3200 (Central Europe)	1:50(Finland), 1:160 (Central Europe)
OTC	Ornithine transcarbamylase deficiency	Ornithine transcarbamylase deficiency is an inherited disorder caused by mutations in the OTC gene. The OTC gene provides instructions for making the enzyme ornithine transcarbamylase. This enzyme participates in the urea cycle and without this enzyme, excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. Ammonia is especially damaging to the nervous system. Other symptoms are lethargy, seizures, coma, development dealy, and intellectual disability. Liver damage may also occur. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:20000 (Central Europe, for women) and the risk of birth of an affected child is lower than 1:80000.	<1:200000	1:20000 females (Central Europe)

PAH	Phenylketonuria	Phenylketonuria is an inherited metabolic disorder caused by mutations in the PAH gene. The PAH gene provides instructions for making an enzyme called phenylalanine hydroxylase. This enzyme converts phenylalanine to other important compounds in the body. Due to a mutations in the PAH gene, phenylalanine from the diet is not processed effectively and can build up to toxic levels in the blood and other tissues instead. Nerve cells in the brain are particularly sensitive to phenylalanine. Without treatment, children can develop intellectual disability, seizures, delayed development, behavioral problems, and psychiatric disorders. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1330 (Central Europe) and the risk of birth of an affected child is lower than 1:5320.	<1:1330	1:40 (Central Europe)
PCDH15	Usher syndrome type 1D	Usher syndrome type 1D is a condition caused by mutations in the PCDH15 gene. Mutations in the PCDH15 gene affect sensory cells in the inner ear and retina. Degeneration of these sensory cells causes the hearing loss, balance and coordination problems, and vision loss. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:750 (Central Europe) and the risk of birth of an affected child is lower than 1:3000.	<1:750	1:450 (Central Europe)
PEX1	Zellweger syndrome type 1A	Zellweger syndrome type 1A is a disorder caused by mutations in the PEX1 gene. The PEX1 gene provides instructions for making a protein called peroxisomal biogenesis factor 1 (Pex1p) from a group of proteins called peroxins, which are essential for the formation and normal functioning of peroxisomes. Peroxisomes contain enzymes needed to break down many different substances. Mutations in the PEX1 gene prevent peroxisomes from functioning normally. Signs and symptoms of Zellweger syndrome are hypotonia, hearing and vision loss, and seizures, all caused by the breakdown of myelin. Other signs are problems with liver, heart, kidney, skeletal abnormalities, and distinctive facial features. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2800 (Central Europe) and the risk of birth of an affected child is lower than 1:11200.	<1:2800	1:140 (Central Europe)
PEX12	Zellweger syndrome type 3A	Zellweger syndrome type 3A is a disorder caused by mutations in the PEX12 gene. The PEX12 gene provides instructions for making a protein called Peroxisome assembly protein 12 from a group of proteins called peroxins, which are essential for the formation and normal functioning of peroxisomes. Peroxisomes contain enzymes needed to break down many different substances. Mutations in the PEX12 gene prevent peroxisomes from functioning normally. Signs and symptoms of Zellweger syndrome are hypotonia, hearing and vision loss, and seizures, all caused by the breakdown of myelin. Other signs are problems with liver, heart, kidney, skeletal abnormalities, and distinctive facial features. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2000 (Central Europe) and the risk of birth of an affected child is lower than 1:8000.	<1:2000	1:350 (Central Europe)
PEX6	Zellweger syndrome type 4A	Zellweger syndrome type 4A is a disorder caused by mutations in the PEX6 gene. The PEX6 gene provides instructions for making a protein called Peroxisomal Biogenesis Factor 6 from a group of proteins called peroxins, which are essential for the formation and normal functioning of peroxisomes. Peroxisomes contain enzymes needed to break down many different substances. Mutations in the PEX6 gene prevent peroxisomes from functioning normally. Signs and symptoms of Zellweger syndrome are hypotonia, hearing and vision loss, and seizures, all caused by the breakdown of myelin. Other signs are problems with liver, heart, kidney, skeletal abnormalities, and distinctive facial features. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:5600 (Central Europe) and the risk of birth of an affected child is lower than 1:22400.	<1:5600	1:280 (Central Europe)

PEX7	Rhizomelic chondrodysplasia punctata type 1	Rhizomelic chondrodysplasia punctata type 1 is a disorder caused by mutations in the PEX7 gene. The PEX7 gene provides instructions for making a peroxisomal biogenesis factor 7 from a group of proteins called peroxins, which are essential for the formation and normal functioning of peroxisomes. Within peroxisomes, the proteins produced from the PEX7 genes play roles in the formation of plasmalogens. Mutations in the PEX7 gene prevent peroxisomes from making plasmalogens, causing Rhizomelic chondrodysplasia punctata. Signs and symptoms are skeletal abnormalities, distinctive facial features, intellectual disability, and respiratory problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe) and the risk of birth of an affected child is lower than 1:12800.	<1:3200	1:160 (Central Europe)
PKHD1	Polycystic kidney disease type 4	Polycystic kidney disease type 4 is a disorder caused by mutations in the PKHD1 gene. This gene provides instructions for making a protein fibrocystin, whose exact function is unknown. Mutations in the PKHD1 gene lead to the formation of numerous cysts characteristic of polycystic kidney disease. Grow of cysts causes the kidneys to become enlarged, unable to filter waste products and may also lead to kidney failure. Cysts may also develop in other organs, particularly the liver. Other symptoms are hypertension, hematuria, kidney stones, urinary track infections, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1400 (Central Europe) and the risk of birth of an affected child is lower than 1:5600.	<1:1400	1:70 (Central Europe)
PMM2	Congenital disorder of glycosylation type Ia	Congenital disorder of glycosylation type Ia is an inherited condition caused by mutations in the PMM2 gene. The PMM2 gene provides instructions for making an enzyme called phosphomannomutase 2, which is involved in the glycosylation, a process of attaching oligosaccharides to proteins. Mutations in the PMM2 gene lead to the production of an abnormal PMM2 enzyme and incorrect oligosaccharides are attached to proteins. Symptoms of the condition are psychomotor retardation, seizures, abnormal distribution of fat, underdeveloped cerebellum, and multivisceral damage. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1400 (Central Europe) and the risk of birth of an affected child is lower than 1:5600.	<1:1400	1:70 (Central Europe)
POLG	Progressive external ophthalmoplegia, Mitochondrial DNA depletion syndrome 4A and 4B, Mitochondrial ataxia syndrome	The POLG gene provides instructions for making the alpha subunit of a protein called polymerase gamma (Pol γ). Pol γ is found in mitochondria and is important for replicating cell´s genetic material and it also plays roles in DNA repair. Mutations in the POLG gene are related to several health conditions: progressive external ophthalmoplegia, mitochondrial DNA depletion syndrome 4A and 4B, and mitochondrial ataxia syndrome. Signs and symptoms of these conditions are linked to abnormal function of Pol γ and defects in DNA synthesis and repair. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:2260 (Central Europe) and the risk of birth of an affected child is lower than 1:9040.	<1:2260	1:113 (Central Europe)
PPT1	Neuronal ceroid lipofuscinosis type 1	Neuronal ceroid lipofuscinosis type 1 is a condition caused by mutations in the PPT1 gene. The PPT1 gene provides instructions for making an enzyme called palmitoyl-protein thioesterase 1, found in the lysosomes. This enzyme breaks down specific proteins and PPT1 gene mutations reduce its function. Partially broken down fats and proteins accumulate in lysosomes, damaging the nerve cells. Symptoms of this condition are development regression, loss of brain tissue, microcephaly, epilepsy, and intellectual disability. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1800 (Central Europe, 1:1200 for Scandinavia) and the risk of birth of an affected child is lower than 1:7200 (1:4800 fo Scandinavia).	<1:1200 (Scandinavia), <1:1800 (Central Europe)	1:60 (Scandinavia), 1:90 (Central Europe)

PROP1	Combined pituitary hormone deficiency	Combined pituitary hormone deficiency is a condition caused by mutations in the PROP1 gene. The PROP1 gene provide instructions for making proteins called transcription factors, are involved in the development of the pituitary gland. Mutations in the PROP1 gene can result in abnormal differentiation of pituitary gland cells and may prevent the production of several hormones, such as cortisol. Symptoms of this condition are delayed or absent puberty, infertility, an unactivity of thyroid gland. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:900 (worldwide) and the risk of birth of an affected child is lower than 1:3600.	<1:900	1:45 (worldwide)
RNASEH2B	Aicardi-Goutieres syndrome	Aicardi-Goutières syndrome is a disorder caused by mutations in the RNASEH2B gene. The RNASEH2B gene provides instructions for making one part of the RNase H2 complex that helps break down molecules containing RNA. Gene mutations result in absence or abnormal function of the complex. The excess RNA may be mistaken by cells for the genetic material of viral invaders, triggering immune system reactions. Signs of Aicardi-Goutières syndrome are brain dysfunction (encephalopathy), skin lesions, and other health problems associated with abnormal immune system activation. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1600 (worldwide) and the risk of birth of an affected child is lower than 1:6400.	<1:1600	1:160 (worldwide)
SBDS	Shwachman-Diamond syndrome	Shwachman-Diamond syndrome is an inherited condition caused by mutations in the SBDS gene. The SBDS encodes a protein that is critical for building ribosomes. Gene mutations impair the formation of ribosomes, which may reduce the production of other proteins and alter developmental processes. This affects many parts of the body, particularly the bone marrow, pancreas, and bones. Some symptoms of Shwachman-Diamond syndrome are bone marrow malfunctions, anemia, infections of the lungs, ears, or skin. Acute myeloid leukemia is also common. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1350 (worldwide) and the risk of birth of an affected child is lower than 1:5400.	<1:1350	1:135 (worldwide)
SERPINA1	Alpha-1 antitrypsin deficiency	Alpha-1 antitrypsin deficiency is an inherited disorder caused by mutations in the SERPINA1 gene. The SERPINA1 encodes a protein called alpha-1 antitrypsin, which protects the body from an enzyme called neutrophil elastase. Neutrophil elastase is released from white blood cells to fight infection, but it can attack normal tissues if not controlled by alpha-1 antitrypsin. Mutations in the SERPINA1 gene can lead to a deficiency of alpha-1 antitrypsin and neutrophil elastase destroys alveoli and causes lung disease. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:800 (Central Europe) and the risk of birth of an affected child is lower than 1:3200.	<1:800	1:40 (Central Europe)
SGSH	Mucopolysaccharidosis type IIIA	Mucopolysaccharidosis type IIIA is a progressive disorder caused by mutations in the SGSH gene. The SGSH gene provides instructions for producing a sulfamidase enzyme that is involved in the breakdown of heparan sulfate. Mutations in the SGSH gene reduce or eliminate the function of the enzyme and the breakdown of a heparan sulfate is disrupted. As a result, partially broken down heparan sulfate accumulates within lysosomes, which negatively affects the brain and spinal cord, causing neurodegeneration. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe) and the risk of birth of an affected child is lower than 1:12000.	<1:3000	1:150 (Central Europe)

SLC26A2	Achondrogenesis type Ib, Atelosteogenesis type II, Diastrophic dysplasia, Multiple epiphyseal dysplasia	The SLC26A2 gene provides instructions for making a protein that transports ions across cell membranes. This protein appears to be active in many of the body's tissues, including developing cartilage, which is a tissue that is later converted to bone. Mutations in the SLC26A2 gene prevent the production of functional protein, which affects the structure of cartilage and the normal formation and growth of bones. Mutations are related to several health conditions with disrupted bone development: achondrogenesis type Ib, atelosteogenesis type II, diastrophic dysplasia, and multiple epiphyseal dysplasia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3000 (Central Europe) and the risk of birth of an affected child is lower than 1:12000.	<1:3000	1:150 (Central Europe)
SLC26A4	Pendred syndrome, Deafness, autosomal recessive 4	The SLC26A4 gene provides instructions for making a protein called pendrin that transports cations across cell membranes. Pendrin is produced in several organs and tissues, particularly the inner ear and thyroid gland. Mutations in the SLC26A4 gene lead to Pendred syndrome, or deafness. Mutations impair or eliminate the activity of pendrin, which disrupts ion transport. In the inner ear, the development of the cochlea and vestibular aqueduct is impaired and the changes in ion levels also lead to the loss of sensory cells that are needed for hearing. In the thyroid, iodide ions are not available for thyroid hormone production and in order to compensate the lack of iodide, the thyroid tissue enlarges. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1600 (Central Europe) and the risk of birth of an affected child is lower than 1:6400.	<1:1600	1:80 (Central Europe)
SLC37A4	Glycogen storage disease type Ib, Ic	Glycogen storage disease type Ib and Ic is an inherited disorder caused by mutations in the SLC37A4 gene. Proteins produced from the SLC37A4 gene break down a glucose 6-phosphate into a glucose. Due to a mutations in G6PC gene, glucose 6-phosphate is not broken down into glucose, but converted to glycogen and fat, so it can be stored within cells. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys. Symptoms of this condition are enlarged liver and kidneys, xanthomas, osteoporosis, arthritis, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe) and the risk of birth of an affected child is lower than 1:12800.	<1:3200	1:160 (Central Europe)
SMN1	Spinal muscular atrofy	Spinal muscular atrophy is a genetic disorder caused by mutations in the SMN1 gene. The SMN1 gene provides instructions for making the SMN protein, which is important for the maintenance of motor neurons in the spinal cord and the brainstem. Motor neurons transmit signals from the brain and spinal cord that tell skeletal muscles to contract, which allows the body to move. Mutations in the SMN1 gene lead to shortage of SMN protein and motor neuron death. Muscles cannot contract without receiving signals from the brain, so many skeletal muscles become weak and waste away. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1000 (Central Europe) and the risk of birth of an affected child is lower than 1:4000.	<1:1000	1:50 (Central Europe)
SMPD1	Niemann-Pick disease	Niemann-Pick disease is a condition caused by mutations in the SMPD1 gene, encoding an enzyme called acid sphingomyelinase. Acid sphingomyelinase is responsible for the conversion of a fat called sphingomyelin into ceramide. Mutations in SMPD1 lead to a shortage of acid sphingomyelinase, causing a sphingomyelin to accumulate in cells. This fat buildup causes cells to malfunction and eventually die, which impairs function of tissues and organs including the brain, lungs, spleen, and liver. Symptoms of the condition are enlarged liver and spleen, loss of mental abilities and movement, lung infections, an eye abnormality, and others. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:4800 (Central Europe) and the risk of birth of an affected child is lower than 1:19200.	<1:4800	1:240 (Central Europe)

TGM1	Congenital ichthyosis	Congenital ichthyosis is a condition caused by mutations in the TGM1 gene. TGM1 gene encodes transglutaminase-1, a catalytic membrane-bound enzyme that functions in the formation of the epidermal cornified cell envelope. TGM1 gene mutations lead to severely reduced or absent enzyme production, which prevents the formation of the cornified cell envelope. Clinical symptoms in affected patients are skin scaling over the whole body, collodion membrane at birth with ectropion and eclabium, erythema, hypohidrosis, palmoplantar hyperkeratosis, dystrophic nails, joint contractures, alopecia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3100 (Central Europe) and the risk of birth of an affected child is lower than 1:12400.	<1:3100	1:155 (Central Europe)
TMEM216	Joubert syndrome type 2 Meckel syndrome type 2	Meckel and Joubert syndrome type 2 are conditions associated with mutations in the TMEM216 gene. Mutations lead to problems with the structure and function of primary cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells and are involved in sensing the physical environment and in chemical signaling. Defects in these cell structures disrupt important chemical signaling pathways during early development. Symptoms of Meckel and Joubert syndrome are abnormalities of the brain and spinal cord, polydactyly, problems with development of the eyes and other facial features, heart, bones, urinary system, and genitalia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1840 (Ashkenazi) and the risk of birth of an affected child is lower than 1:7360.	<1840	1:92 (Ashkenazi)
TPP1	Neuronal ceroid lipofuscinosis type 2 Spinocerebellar ataxia type 7	The TPP1 gene provides instructions for making an enzyme called tripeptidyl peptidase 1. This enzyme breaks down peptides into amino acids. Mutation in the TPP1 disrupt the break down of peptides and they can lead to a conditions know as neuronal ceroid lipofuscinosis type 2 and spinocerebellar ataxia type 7. They are both neurodegenerative disorders characterized by dementia, seizures, visual loss or eye abnormalities, and other neurological problems. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (Central Europe) and the risk of birth of an affected child is lower than 1:12800.	<1:3200	1:160 (Central Europe)
TSHR	Congenital hypothyroidism	Congenital hypothyroidism is a condition caused by mutations in the TSHR gene. Mutations in the TSHR gene lead to interruption of interaction with thyroid stimulating hormone. Therefore, thyroid hormone production is not stimulated. Congenital hypothyroidism is characterised by abnormally low levels of thyroid hormones starting from birth. Other symptoms are lessactivity in newborn, difficulty feeding, constipation and if untreated intelectual disability and growth restriction. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (worldwide) and the risk of birth of an affected child is lower than 1:12800.	<1:3200	1:160 (worldwide)
TYR	Oculocutaneous albinism type 1A and 1B	Oculocutaneous albinism type 1A and 1B is a condition caused by mutations in the TYR gene. Tyrosinase encoded by TYR gene catalyzes several steps in conversion of tyrosine to melanin. This enzyme is located in melanocytes. Mutation in the TYR gene reduce or eliminate the activity of tyrosinase, preventing melanocytes from producing melanin. Affected individuals have hypopigmentation of the skin, eyes and hair, reduced visual acuity, nystagmus, strabism and foveal hypoplasia. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:1420 (worldwide) and the risk of birth of an affected child is lower than 1:5680.	<1:1420	1:71 (worldwide)

USH2A	Usher syndrome type 2A	Usher syndrome type 2A is a condition caused by mutations in the USH2A gene. The USH2A gene provide instructions for making protein caled usherin. Usherin is found in basement membranes in the inner ear and in the retina. Most of the gene mutations responsible for Usher syndrome lead to a loss of hair cells in the inner ear and a gradual loss of rods and cones in the retina. Degeneration of these sensory cells hearing loss, vision loss, and problems with balance and coordination. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:800 (Central Europe) and the risk of birth of an affected child is lower than 1:3200.	<1:800	1:80 (Central Europe)
USH1C	Usher syndrome type 1C	Usher syndrome type 1C is a condition caused by mutations in the USH1C gene. This gene encodes a scaffold protein that functions in the assembly of Usher protein complexex that functions in the inner ear and in the retina. Most of the gene mutations responsible for Usher syndrome lead to a loss of hair cells in the inner ear and a gradual loss of rods and cones in the retina. Degeneration of these sensory cells hearing loss, vision loss, and problems with balance and coordination. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:700 (Central Europe) and the risk of birth of an affected child is lower than 1:2800.	<1:700	1:350 (Central Europe)
XPC	Xeroderma pigmentosum	Xeroderma pigmentosum is a condition caused by mutations in the XPC gene. The XPC gene provides instructions for making a protein that is involved in a DNA-repair process known as nucleotide excision repair. DNA can be damaged by UV rays and cells with mutations in the XPC gene are not able to repair this damage. Instead, unrepaired DNA is accumulated in the cells. Xeroderma pigmentosum is characterized by an extreme sensitivity to ultraviolet rays from sunlight, particulary in areas of skin exposed to the sun and eyes. DNA damage in genes that control cell growth and division can lead to the development of cancerous tumors. In case of negative result using the same test in partner, the residual risk of disease transmission is lower than 1:3200 (worldwide) and the risk of birth of an affected child is lower than 1:12800.	<1:3200	1:160 (worldwide)