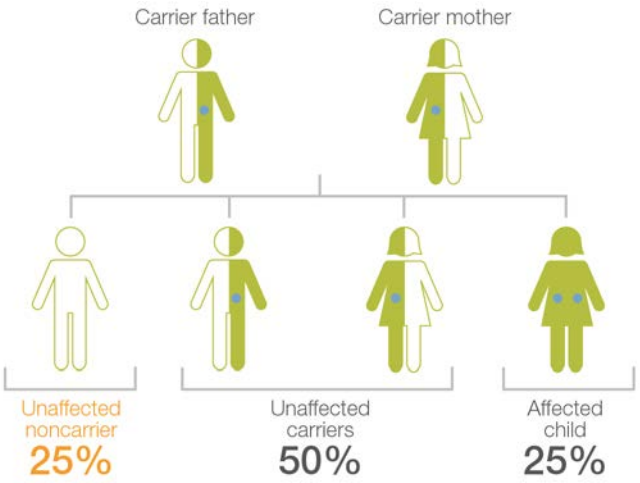
genescreen®

## About GeneScreen® Easy - Carrier Screening Test

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

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




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

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

## Carrier testing with comprehensive coverage

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

Using NGS technique, **GeneScreen® Easy** Carrier Screening Test screens **330 genes** for mutations associated with **over 500** recessive and X-linked **genetic disorders**, including the most common in the European population**.** This provides patients more opportunities to identify potential hereditary risks.

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

## Carrier screen with unmatched detection of serious disorders

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

The **GeneScreen® Easy** test screens for the most clinically relevant and impactful genetic conditions



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

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

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

## Knowing Enables Informed Decisions

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

## GeneScreen® Easy: Indication for testing

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

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

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



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

to avoid high reproductive risk pregnancies.
- Anyone who wants to know if they are carrier of any condition included in the panel.

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

## GeneScreen® Easy: The Testing Process

The DNA is first isolated from the peripheral blood and then **amplified by PCR.** Through a state-of-the-art technological process, named **massively parallel sequencing** (**MPS**), which uses ***Next Generation Sequencing*** (**NGS**) techniques with **ILLUMINA** sequencing instruments, **330 genes** are completely sequenced (whole exons sequencing, including adjacent intronic regions, ± 5 nucleotides) (Table 1) at high read depth. The resulting genetic sequences are analysed via an **advanced bioinformatics analysis**, to assess the presence of potential mutations in the genes under investigation.
**GeneScreen® Easy** test, unlike other carrier screening tests using targeted sequencing, performs **full-exon sequencing** of all the genes included on the panel, which allows a more comprehensive analysis of each gene and related diseases. Gene dosage analysis by Multiplex Ligation-dependent Probe Amplification (MLPA) of the SMN1 gene was performed for SMA carrier screening. Fluorescent PCR was used for Fragile-X carrier screen, to detect the (CGG)n repeat expansions in the promoter region of the FMR-1 gene.

## Results of the GeneScreen® Easy test

**"POSITIVE" – Presence of one or more mutations:** this result shows that the test detected one or more mutations in one or more genes, identifying the patient as a **carrier**. Mutations reported through the **GeneScreen® Easy** test may be classified under the following prognosis categories:

- **Known pathogenic**: clinical relevant mutations causing well-established syndromes;
- **Likely pathogenic**: variants that are likely clinical relevant and may cause well-established syndromes.
- **Variants of uncertain clinical significance (VOUS):** findings with insufficient evidence available for unequivocal determination of clinical significance.

Classification follows the recommendations of the international reference guidelines (Richards et al., 2015)
Being a carrier is relatively common, carriers do not generally involve the disease development and do not show any symptoms. A patient with a positive test result should be referred for genetic counseling and further evaluation. The patient's reproductive partner and at-risk family members may also be tested. When both parents are carrier of the same genetic disease, there is a 25% chance of transmitting this condition to their offspring. In these cases, genetic counseling is recommended to the couple, where various reproductive choices will be discussed, including:

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

**"NEGATIVE" - No mutations:** this result shows the test has not detected any disease causing mutation in the investigated genes.



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

## Parameters used to report the genetic variations

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

## Target Coverage

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

## Accuracy of the GeneScreen® Easy test

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

## Limitation of the GeneScreen® Easy test

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

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

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



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

geneticists to have a better interpretation of genetic results.
The intrinsic limitation of the NGS methodology is the lack of coverage uniformity of each examined genetic region. Quantity and quality of the DNA extracted from prenatal samples is one of the potential causes of such lack of uniformity, which may lead to the lack of detection of gene mutations. Due to this limitation, NGS tests may not detect specific genetic mutations in the selected genes.

## Genetic Counseling

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

## References

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine. 2015;17(5):405-423.
2. Henneman L, Borry P, Chokoslivi D, et al. Responsible implementation of expanded carrier screening. European Journal of Human Genetics. 2016;24,e1–e12.
3. Edwards JG, Feldman G, Goldberg J, et al. Expanded Carrier Screening in Reproductive Medicine-points to Consider. A Joint Statement of the American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine. Obstet Gynecol 2015;125(3):653–662.
4. Haque IS, Lazarin GA, Kang HP, et al. Modeled Fetal Risk of Genetic Diseases Identified by Expanded Carrier Screening. JAMA. 2016;316(7):734-742.



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

**Table 1: GeneScreen® Easy - List of genes screened and genetic diseases investigated**

| Gene | OMIM Gene | Disease | OMIM Disease | Inheritance |
|---|---|---|---|---|
| ABCD1 | 300371 | Adrenoleukodystrophy | 300100 | XLR |
| ABCC8 | 600509 | Diabetes mellitus, noninsulin-dependent | 125853 | AD |
| | | Diabetes mellitus, permanent neonatal | 606176 | AD, AR |
| | | Diabetes mellitus, transient neonatal 2 | 610374 | |
| | | Hyperinsulinemic hypoglycemia, familial, 1 | 256450 | AD, AR |
| | | Hypoglycemia of infancy, leucine-sensitive | 240800 | AD |
| ABCB11 | 603201 | Cholestasis, benign recurrent intrahepatic, 2 | 605479 | AR |
| | | Cholestasis, progressive familial intrahepatic 2 | 601847 | AR |
| ADAMTS2 | 604539 | Ehlers-Danlos syndrome, dermatosparaxis type | 225410 | AR |
| EVC | 604831 | ?Weyers acrofacial dysostosis | 193530 | AD |
| ACADS | 606885 | Acyl-CoA dehydrogenase, short-chain, deficiency of | 201470 | AR |
| ACADM | 607008 | Acyl-CoA dehydrogenase, medium chain, deficiency of | 201450 | AR |
| EVC2 | 607261 | Ellis-van Creveld syndrome | 225500 | AR |
| ACAT1 | 607809 | Alpha-methylacetoacetic aciduria | 203750 | AR |
| ADA | 608958 | Adenosine deaminase deficiency, partial | 102700 | SMo, AR |
| ACADVL | 609575 | VLCAD deficiency | 201475 | AR |
| ACOX1 | 609751 | Peroxisomal acyl-CoA oxidase deficiency | 264470 | AR |
| AGL | 610860 | Glycogen storage disease IIIa | 232400 | AR |
| ACAD9 | 611103 | Mitochondrial complex I deficiency, nuclear type 20 | 611126 | AR |
| AGA | 613228 | Aspartylglucosaminuria | 208400 | AR |
| | | Severe combined immunodeficiency due to ADA deficiency | 102700 | SMo, AR |
| | | Glycogen storage disease IIIb | 232400 | AR |
| AGPS | 603051 | Rhizomelic chondrodysplasia punctata, type 3 | 600121 | AR |
| AGXT | 604285 | Hyperoxaluria, primary, type 1 | 259900 | AR |
| AIRE | 607358 | Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia | 240300 | AD, AR |
| ALDH3A2 | 609523 | Sjogren-Larsson syndrome | 270200 | AR |
| ALDOB | 612724 | Fructose intolerance, hereditary | 229600 | AR |
| ALG6 | 604566 | Congenital disorder of glycosylation, type Ic | 603147 | AR |
| ALMS1 | 606844 | Alstrom syndrome | 203800 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| ALPL | 171760 | Hypophosphatasia, adult | 146300 | AD, AR |
| | | Hypophosphatasia, childhood | 241510 | AR |
| | | Hypophosphatasia, infantile | 241500 | AR |
| | | Odontohypophosphatasia | 146300 | AD, AR |
| AMT | 238310 | Glycine encephalopathy | 605899 | AR |
| AR | 313700 | Androgen insensitivity | 300068 | XLR |
| | | Androgen insensitivity, partial, with or without breast cancer | 312300 | XLR |
| | | Hypospadias 1, X-linked | 300633 | XLR |
| | | Spinal and bulbar muscular atrophy of Kennedy | 313200 | XLR |
| | | Prostate cancer, susceptibility to | 176807 | AD, SMu |
| ARG1 | 608313 | Argininemia | 207800 | AR |
| ARSA | 607574 | Metachromatic leukodystrophy | 250100 | AR |
| ARSB | 611542 | Mucopolysaccharidosis type VI (Maroteaux-Lamy) | 253200 | AR |
| ARSE | 300180 | Chondrodysplasia punctata, X-linked recessive | 302950 | XLR |
| ASL | 608310 | Argininosuccinic aciduria | 207900 | AR |
| ASNS | 108370 | Asparagine synthetase deficiency | 615574 | AR |
| ASPA | 608034 | Canavan disease | 271900 | AR |
| ASS1 | 603470 | Citrullinemia | 215700 | AR |
| ATM | 607585 | Ataxia-telangiectasia | 208900 | AR |
| | | Lymphoma, B-cell non-Hodgkin, somatic | | |
| | | Lymphoma, mantle cell, somatic | | |
| | | T-cell prolymphocytic leukemia, somatic | | |
| | | Breast cancer, susceptibility to | 114480 | AD, SMu |
| ATP6V1B1 | 192132 | Renal tubular acidosis with deafness | 267300 | AR |
| ATP7A | 300011 | Menkes disease | 309400 | XLR |
| | | Occipital horn syndrome | 304150 | XLR |
| | | Spinal muscular atrophy, distal, X-linked 3 | 300489 | XLR |
| ATP7B | 606882 | Wilson disease | 277900 | AR |
| ATRX | 300032 | Alpha-thalassemia myelodysplasia syndrome, somatic | 300448 | |
| | | Alpha-thalassemia/mental retardation syndrome | 301040 | XLD |
| | | Mental retardation-hypotonic facies syndrome, X-linked | 309580 | XLR |
| BBS1 | 209901 | Bardet-Biedl syndrome 1 | 209900 | AR, DR |
| BBS10 | 610148 | Bardet-Biedl syndrome 10 | 615987 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| BBS12 | 610683 | Bardet-Biedl syndrome 12 | 615989 | AR |
| BBS2 | 606151 | Bardet-Biedl syndrome 2 | 615981 | AR |
| | | Retinitis pigmentosa 74 | 616562 | AR |
| BCHE | 177400 | Butyrylcholinesterase deficiency | 617936 | |
| | | Apnea, postanesthetic, susceptibility to, due to BCHE deficiency | 617936 | |
| BCKDHA | 608348 | Maple syrup urine disease, type Ia | 248600 | AR |
| BCKDHB | 248611 | Maple syrup urine disease, type Ib | 248600 | AR |
| BCS1L | 603647 | Bjornstad syndrome | 262000 | AR |
| | | GRACILE syndrome | 603358 | |
| | | Leigh syndrome | 256000 | Mi, AR |
| | | Mitochondrial complex III deficiency, nuclear type 1 | 124000 | AR |
| BLM | 604610 | Bloom syndrome | 210900 | AR |
| BSND | 606412 | Bartter syndrome, type 4a | 602522 | AR |
| | | Sensorineural deafness with mild renal dysfunction | 602522 | AR |
| BTD | 609019 | Biotinidase deficiency | 253260 | AR |
| BTK | 300300 | Agammaglobulinemia, X-linked 1 | 300755 | XLR |
| | | Isolated growth hormone deficiency, type III, with agammaglobulinemia | 307200 | XLR |
| CAPN3 | 114240 | Muscular dystrophy, limb-girdle, autosomal dominant 4 | 618129 | AD |
| | | Muscular dystrophy, limb-girdle, autosomal recessive 1 | 253600 | AR |
| CBS | 613381 | Homocystinuria, B6-responsive and nonresponsive types | 236200 | AR |
| | | Thrombosis, hyperhomocysteinemic | 236200 | AR |
| CC2D2A | 612013 | COACH syndrome | 216360 | AR |
| | | Joubert syndrome 9 | 612285 | AR |
| | | Meckel syndrome 6 | 612284 | AR |
| CD40LG | 300386 | Immunodeficiency, X-linked, with hyper-IgM | 308230 | XLR |
| CDH23 | 605516 | Deafness, autosomal recessive 12 | 601386 | AR |
| | | Usher syndrome, type 1D | 601067 | AR, DR |
| | | Usher syndrome, type 1D/F digenic | 601067 | AR, DR |
| | | Pituitary adenoma 5, multiple types | 617540 | AD |
| CEP290 | 610142 | Bardet-Biedl syndrome 14 | 615991 | AR |
| | | Joubert syndrome 5 | 610188 | AR |
| | | Leber congenital amaurosis 10 | 611755 | |
| | | Meckel syndrome 4 | 611134 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Senior-Loken syndrome 6 | 610189 | AR |
| CERKL | 608381 | Retinitis pigmentosa 26 | 608380 | |
| CFTR | 602421 | Congenital bilateral absence of vas deferens | 277180 | AR |
| | | Cystic fibrosis | 219700 | AR |
| | | Sweat chloride elevation without CF | | |
| | | Bronchiectasis with or without elevated sweat chloride 1, modifier of | 211400 | AD |
| | | Hypertrypsinemia, neonatal | | |
| | | Pancreatitis, hereditary | 167800 | AD |
| CHM | 300390 | Choroideremia | 303100 | XLD |
| CHRNE | 100725 | Myasthenic syndrome, congenital, 4A, slow-channel | 605809 | AD, AR |
| | | Myasthenic syndrome, congenital, 4B, fast-channel | 616324 | AR |
| | | Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency | 608931 | AR |
| CLN3 | 607042 | Ceroid lipofuscinosis, neuronal, 3 | 204200 | AR |
| CLN5 | 608102 | Ceroid lipofuscinosis, neuronal, 5 | 256731 | AR |
| CLN6 | 606725 | Ceroid lipofuscinosis, neuronal, 6 | 601780 | AR |
| | | Ceroid lipofuscinosis, neuronal, Kufs type, adult onset | 204300 | AR |
| CLN8 | 607837 | Ceroid lipofuscinosis, neuronal, 8 | 600143 | AR |
| | | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant | 610003 | AR |
| CLRN1 | 606397 | Retinitis pigmentosa 61 | 614180 | |
| | | Usher syndrome, type 3A | 276902 | AR |
| CNGB3 | 605080 | Achromatopsia 3 | 262300 | AR |
| | | Macular degeneration, juvenile | 248200 | AR |
| COL17A1 | 113811 | Epidermolysis bullosa, junctional, localisata variant | 226650 | AR |
| | | Epidermolysis bullosa, junctional, non-Herlitz type | 226650 | AR |
| | | Epithelial recurrent erosion dystrophy | 122400 | AD |
| COL4A3 | 120070 | Alport syndrome 2, autosomal recessive | 203780 | AR |
| | | Alport syndrome 3, autosomal dominant | 104200 | AD |
| | | Hematuria, benign familial | 141200 | AD |
| COL4A4 | 120131 | Alport syndrome 2, autosomal recessive | 203780 | AR |
| | | Hematuria, familial benign | 141200 | AD |
| COL4A5 | 303630 | Alport syndrome 1, X-linked | 301050 | XLD |
| COL7A1 | 120120 | EBD inversa | 226600 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | EBD, Bart type | 132000 | AD |
| | | EBD, localisata variant | | |
| | | Epidermolysis bullosa dystrophica, AD | 131750 | AD |
| | | Epidermolysis bullosa dystrophica, AR | 226600 | AR |
| | | Epidermolysis bullosa pruriginosa | 604129 | AD, AR |
| | | Epidermolysis bullosa, pretibial | 131850 | AD, AR |
| | | Toenail dystrophy, isolated | 607523 | AD |
| | | Transient bullous of the newborn | 131705 | AD, AR |
| CPS1 | 608307 | Carbamoylphosphate synthetase I deficiency | 237300 | AR |
| | | Pulmonary hypertension, neonatal, susceptibility to | 615371 | |
| | | Venoocclusive disease after bone marrow transplantation | | |
| CPT1A | 600528 | CPT deficiency, hepatic, type IA | 255120 | AR |
| CPT2 | 600650 | CPT II deficiency, infantile | 600649 | AR |
| | | CPT II deficiency, lethal neonatal | 608836 | AR |
| | | CPT II deficiency, myopathic, stress-induced | 255110 | AD, AR |
| | | Encephalopathy, acute, infection-induced, 4, susceptibility to | 614212 | AD, AR |
| CRB1 | 604210 | Leber congenital amaurosis 8 | 613835 | |
| | | Pigmented paravenous chorioretinal atrophy | 172870 | AD |
| | | Retinitis pigmentosa-12 | 600105 | AR |
| CRTAP | 605497 | Osteogenesis imperfecta, type VII | 610682 | AR |
| CSTB | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) | 254800 | AR |
| CTNS | 606272 | Cystinosis, atypical nephropathic | 219800 | AR |
| | | Cystinosis, late-onset juvenile or adolescent nephropathic | 219900 | AR |
| | | Cystinosis, nephropathic | 219800 | AR |
| | | Cystinosis, ocular nonnephropathic | 219750 | AR |
| CTSD | 116840 | Ceroid lipofuscinosis, neuronal, 10 | 610127 | AR |
| CTSK | 601105 | Pycnodysostosis | 265800 | AR |
| CYP11B1 | 610613 | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency | 202010 | AR |
| | | Aldosteronism, glucocorticoid-remediable | 103900 | AD |
| CYP11B2 | 124080 | Aldosterone to renin ratio raised | | |
| | | Hypoaldosteronism, congenital, due to CMO I deficiency | 203400 | AR |
| | | Hypoaldosteronism, congenital, due to CMO II deficiency | 610600 | AR |
| | | Low renin hypertension, susceptibility to | | |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| CYP17A1 | 609300 | 17,20-lyase deficiency, isolated | 202110 | AR |
| | | 17-alpha-hydroxylase/17,20-lyase deficiency | 202110 | AR |
| CYP19A1 | 107910 | Aromatase deficiency | 613546 | |
| | | Aromatase excess syndrome | 139300 | AD |
| CYP1B1 | 601771 | Anterior segment dysgenesis 6, multiple subtypes | 617315 | |
| | | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset | 231300 | AR |
| CYP21A2 | 613815 | Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency | 201910 | AR |
| | | Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency | 201910 | AR |
| CYP27A1 | 606530 | Cerebrotendinous xanthomatosis | 213700 | AR |
| DBT | 248610 | Maple syrup urine disease, type II | 248600 | AR |
| DCLRE1C | 605988 | Omenn syndrome | 603554 | AR |
| | | Severe combined immunodeficiency, Athabascan type | 602450 | AR |
| DHCR7 | 602858 | Smith-Lemli-Opitz syndrome | 270400 | AR |
| DHDDS | 608172 | Congenital disorder of glycosylation, type 1bb | 613861 | AR |
| | | Developmental delay and seizures with or without movement abnormalities | 617836 | AD |
| | | Retinitis pigmentosa 59 | 613861 | AR |
| DKC1 | 300126 | Dyskeratosis congenita, X-linked | 305000 | XLR |
| DLD | 238331 | Dihydrolipoamide dehydrogenase deficiency | 246900 | AR |
| DMD | 300377 | Becker muscular dystrophy | 300376 | XLR |
| | | Cardiomyopathy, dilated, 3B | 302045 | XL |
| | | Duchenne muscular dystrophy | 310200 | XLR |
| DNAH5 | 603335 | Ciliary dyskinesia, primary, 3, with or without situs inversus | 608644 | |
| DNAI1 | 604366 | Ciliary dyskinesia, primary, 1, with or without situs inversus | 244400 | AR |
| DNAI2 | 605483 | Ciliary dyskinesia, primary, 9, with or without situs inversus | 612444 | |
| DOK7 | 610285 | Fetal akinesia deformation sequence | 208150 | AR |
| | | Myasthenic syndrome, congenital, 10 | 254300 | AR |
| DPYD | 612779 | 5-fluorouracil toxicity | 274270 | AR |
| | | Dihydropyrimidine dehydrogenase deficiency | 274270 | AR |
| DYSF | 603009 | Miyoshi muscular dystrophy 1 | 254130 | AR |
| | | Muscular dystrophy, limb-girdle, autosomal recessive 2 | 253601 | AR |
| | | Myopathy, distal, with anterior tibial onset | 606768 | AR |
| EDA | 300451 | Ectodermal dysplasia 1, hypohidrotic, X-linked | 305100 | XLR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Tooth agenesis, selective, X-linked 1 | 313500 | XLD |
| EDAR | 604095 | Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant | 129490 | AD |
| | | Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive | 224900 | AR |
| | | Hair morphology 1, hair thickness | 612630 | |
| EIF2AK3 | 604032 | Wolcott-Rallison syndrome | 226980 | AR |
| EIF2B1 | 606686 | Leukoencephalopathy with vanishing white matter | 603896 | AR |
| EIF2B2 | 606454 | Leukoencephalopathy with vanishing white matter | 603896 | AR |
| | | Ovarioleukodystrophy | 603896 | AR |
| EIF2B3 | 606273 | Leukoencephalopathy with vanishing white matter | 603896 | AR |
| EIF2B4 | 606687 | Leukoencephalopathy with vanishing white matter | 603896 | AR |
| | | Ovarioleukodystrophy | 603896 | AR |
| EIF2B5 | 603945 | Leukoencephalopathy with vanishing white matter | 603896 | AR |
| | | Ovarioleukodystrophy | 603896 | AR |
| EMD | 300384 | Emery-Dreifuss muscular dystrophy 1, X-linked | 310300 | XLR |
| ERCC6 | 609413 | Cerebrooculofacioskeletal syndrome 1 | 214150 | AR |
| | | Cockayne syndrome, type B | 133540 | AR |
| | | De Sanctis-Cacchione syndrome | 278800 | AR |
| | | Premature ovarian failure 11 | 616946 | AD |
| | | UV-sensitive syndrome 1 | 600630 | AR |
| | | Lung cancer, susceptibility to | 211980 | AD, SMu |
| | | Macular degeneration, age-related, susceptibility to, 5 | 613761 | |
| ERCC8 | 609412 | Cockayne syndrome, type A | 216400 | AR |
| | | UV-sensitive syndrome 2 | 614621 | AR |
| ESCO2 | 609353 | Roberts syndrome | 268300 | AR |
| | | SC phocomelia syndrome | 269000 | AR |
| ETFA | 608053 | Glutaric acidemia IIA | 231680 | AR |
| ETFB | 130410 | Glutaric acidemia IIB | 231680 | AR |
| ETFDH | 231675 | Glutaric acidemia IIC | 231680 | AR |
| ETHE1 | 608451 | Ethylmalonic encephalopathy | 602473 | AR |
| EVC2 | 607261 | Ellis-van Creveld syndrome | 225500 | AR |
| | | Weyers acrofacial dysostosis | 193530 | AD |
| EYS | 612424 | Retinitis pigmentosa 25 | 602772 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| F11 | 264900 | Factor XI deficiency, autosomal dominant | 612416 | |
| | | Factor XI deficiency, autosomal recessive | 612416 | |
| F8 | 300841 | Hemophilia A | 306700 | XLR |
| F9 | 300746 | Hemophilia B | 306900 | XLR |
| | | Thrombophilia, X-linked, due to factor IX defect | 300807 | |
| | | Deep venous thrombosis, protection against | 300807 | |
| | | Warfarin sensitivity | 122700 | AD |
| FAH | 613871 | Tyrosinemia, type I | 276700 | AR |
| FANCA | 607139 | Fanconi anemia, complementation group A | 227650 | AR |
| FANCC | 613899 | Fanconi anemia, complementation group C | 227645 | AR |
| FANCG | 602956 | Fanconi anemia, complementation group G | 614082 | |
| FH | 136850 | Fumarase deficiency | 606812 | AR |
| | | Leiomyomatosis and renal cell cancer | 150800 | AD |
| FKRP | 606596 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 | 613153 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5 | 606612 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 | 607155 | AR |
| FKTN | 607440 | Cardiomyopathy, dilated, 1X | 611615 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 | 253800 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4 | 613152 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 | 611588 | AR |
| FMR1 | 309550 | Fragile X syndrome | 300624 | XLD |
| | | Fragile X tremor/ataxia syndrome | 300623 | XLD |
| | | Premature ovarian failure 1 | 311360 | XL |
| G6PC | 613742 | Glycogen storage disease Ia | 232200 | AR |
| G6PD | 305900 | Hemolytic anemia, G6PD deficient (favism) | 300908 | XLD |
| | | Resistance to malaria due to G6PD deficiency | 611162 | |
| GAA | 606800 | Glycogen storage disease II | 232300 | AR |
| GALC | 606890 | Krabbe disease | 245200 | AR |
| GALK1 | 604313 | Galactokinase deficiency with cataracts | 230200 | AR |
| GALNS | 612222 | Mucopolysaccharidosis IVA | 253000 | AR |
| GALT | 606999 | Galactosemia | 230400 | AR |
| GAMT | 601240 | Cerebral creatine deficiency syndrome 2 | 612736 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P.Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| GBA | 606463 | Gaucher disease, perinatal lethal | 608013 | AR |
| | | Gaucher disease, type I | 230800 | AR |
| | | Gaucher disease, type II | 230900 | AR |
| | | Gaucher disease, type III | 231000 | AR |
| | | Gaucher disease, type IIIC | 231005 | AR |
| | | Lewy body dementia, susceptibility to | 127750 | AD |
| | | Parkinson disease, late-onset, susceptibility to | 168600 | Mu, AD |
| GBE1 | 607839 | Glycogen storage disease IV | 232500 | AR |
| | | Polyglucosan body disease, adult form | 263570 | AR |
| GCDH | 608801 | Glutaricaciduria, type I | 231670 | AR |
| GFM1 | 606639 | Combined oxidative phosphorylation deficiency 1 | 609060 | AR |
| GJB1 | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 | 302800 | XLD |
| GJB2 | 121011 | Bart-Pumphrey syndrome | 149200 | AD |
| | | Deafness, autosomal dominant 3A | 601544 | AD |
| | | Deafness, autosomal recessive 1A | 220290 | AR |
| | | Hystrix-like ichthyosis with deafness | 602540 | AD |
| | | Keratitis-ichthyosis-deafness syndrome | 148210 | AD |
| | | Keratoderma, palmoplantar, with deafness | 148350 | AD |
| | | Vohwinkel syndrome | 124500 | AD |
| GJB6 | 604418 | Deafness, autosomal dominant 3B | 612643 | AD |
| | | Deafness, autosomal recessive 1B | 612645 | AR |
| | | Deafness, digenic GJB2/GJB6 | 220290 | AR |
| | | Ectodermal dysplasia 2, Clouston type | 129500 | AD |
| GLA | 300644 | Fabry disease | 301500 | XL |
| | | Fabry disease, cardiac variant | 301500 | XL |
| GLB1 | 611458 | GM1-gangliosidosis, type I | 230500 | AR |
| | | GM1-gangliosidosis, type II | 230600 | AR |
| | | GM1-gangliosidosis, type III | 230650 | AR |
| | | Mucopolysaccharidosis type IVB (Morquio) | 253010 | AR |
| GLDC | 238300 | Glycine encephalopathy | 605899 | AR |
| GLE1 | 603371 | Congenital arthrogryposis with anterior horn cell disease | 611890 | AR |
| | | Lethal congenital contracture syndrome 1 | 253310 | AR |
| GNE | 603824 | Nonaka myopathy | 605820 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Sialuria | 269921 | AD |
| GNPAT | 602744 | Rhizomelic chondrodysplasia punctata, type 2 | 222765 | AR |
| GNPTAB | 607840 | Mucolipidosis II alpha/beta | 252500 | AR |
| | | Mucolipidosis III alpha/beta | 252600 | AR |
| GNS | 607664 | Mucopolysaccharidosis type IIID | 252940 | AR |
| GP9 | 173515 | Bernard-Soulier syndrome, type C | 231200 | AR |
| GRHPR | 604296 | Hyperoxaluria, primary, type II | 260000 | AR |
| GUSB | 611499 | Mucopolysaccharidosis VII | 253220 | AR |
| HADHA | 600890 | Fatty liver, acute, of pregnancy | 609016 | AR |
| | | HELLP syndrome, maternal, of pregnancy | 609016 | AR |
| | | LCHAD deficiency | 609016 | AR |
| | | Trifunctional protein deficiency | 609015 | AR |
| HADHB | 143450 | Trifunctional protein deficiency | 609015 | AR |
| HAX1 | 605998 | Neutropenia, severe congenital 3, autosomal recessive | 610738 | AR |
| HBA1 | 141800 | Erythrocytosis, 7 | 617981 | |
| | | Heinz body anemias, alpha- | 140700 | AD |
| | | Hemoglobin H disease, nondeletional | 613978 | |
| | | Methemoglobinemia, alpha type | 617973 | |
| | | Thalassemias, alpha- | 604131 | |
| HBA2 | 141850 | Erythrocytosis 7 | 617981 | |
| | | Heinz body anemia | 140700 | AD |
| | | Hemoglobin H disease, deletional and nondeletional | 613978 | |
| | | Thalassemia, alpha- | 604131 | |
| HBB | 141900 | Delta-beta thalassemia | 141749 | AD |
| | | Erythrocytosis 6 | 617980 | |
| | | Heinz body anemia | 140700 | AD |
| | | Hereditary persistence of fetal hemoglobin | 141749 | AD |
| | | Methmoglobinemia, beta type | 617971 | |
| | | Sickle cell anemia | 603903 | AR |
| | | Thalassemia, beta | 613985 | |
| | | Thalassemia-beta, dominant inclusion-body | 603902 | |
| | | Malaria, resistance to | 611162 | |
| HEXA | 606869 | GM2-gangliosidosis, several forms | 272800 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Tay-Sachs disease | 272800 | AR |
| | | Hex A pseudodeficiency | 272800 | AR |
| HEXB | 606873 | Sandhoff disease, infantile, juvenile, and adult forms | 268800 | AR |
| HFE | 613609 | Hemochromatosis | 235200 | AR |
| | | Transferrin serum level QTL2 | 614193 | |
| | | Alzheimer disease, susceptibility to | 104300 | AD |
| | | Microvascular complications of diabetes 7 | 612635 | |
| | | Porphyria cutanea tarda, susceptibility to | 176100 | AD, AR |
| | | Porphyria variegata, susceptibility to | 176200 | AD |
| HFE2 | 608374 | Hemochromatosis, type 2A | 602390 | AR |
| HGD | 607474 | Alkaptonuria | 203500 | AR |
| HGSNAT | 610453 | Mucopolysaccharidosis type IIIC (Sanfilippo C) | 252930 | AR |
| | | Retinitis pigmentosa 73 | 616544 | AR |
| HLCS | 609018 | Holocarboxylase synthetase deficiency | 253270 | AR |
| HMGCL | 613898 | HMG-CoA lyase deficiency | 246450 | AR |
| HOGA1 | 613597 | Hyperoxaluria, primary, type III | 613616 | |
| HPS1 | 604982 | Hermansky-Pudlak syndrome 1 | 203300 | AR |
| HPS3 | 606118 | Hermansky-Pudlak syndrome 3 | 614072 | AR |
| HSD17B4 | 601860 | D-bifunctional protein deficiency | 261515 | AR |
| | | Perrault syndrome 1 | 233400 | AR |
| HSD3B2 | 613890 | Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency | 201810 | AR |
| HYLS1 | 610693 | Hydrolethalus syndrome | 236680 | AR |
| IDS | 300823 | Mucopolysaccharidosis II | 309900 | XLR |
| IDUA | 252800 | Mucopolysaccharidosis Ih | 607014 | AR |
| | | Mucopolysaccharidosis Ih/s | 607015 | AR |
| | | Mucopolysaccharidosis Is | 607016 | AR |
| IKBKAP | 603722 | Dysautonomia, familial | 223900 | AR |
| IL2RG | 308380 | Combined immunodeficiency, X-linked, moderate | 312863 | XLR |
| | | Severe combined immunodeficiency, X-linked | 300400 | XLR |
| ISPD | 614631 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 | 614643 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7 | 616052 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| IVD | 607036 | Isovaleric acidemia | 243500 | AR |
| KCNJ11 | 600937 | Diabetes mellitus, transient neonatal, 3 | 610582 | AD |
| | | Diabetes, permanent neonatal, with or without neurologic features | 606176 | AD, AR |
| | | Hyperinsulinemic hypoglycemia, familial, 2 | 601820 | AR |
| | | Maturity-onset diabetes of the young, type 13 | 616329 | AD |
| | | Diabetes mellitus, type 2, susceptibility to | 125853 | AD |
| L1CAM | 308840 | Corpus callosum, partial agenesis of | 304100 | XLR |
| | | CRASH syndrome | 303350 | XLR |
| | | Hydrocephalus due to aqueductal stenosis | 307000 | XLR |
| | | Hydrocephalus with congenital idiopathic intestinal pseudoobstruction | 307000 | XLR |
| | | Hydrocephalus with Hirschsprung disease | 307000 | XLR |
| | | MASA syndrome | 303350 | XLR |
| LAMA2 | 156225 | Muscular dystrophy, congenital, merosin deficient or partially deficient | 607855 | AR |
| | | Muscular dystrophy, limb-girdle, autosomal recessive 23 | 618138 | AR |
| LAMA3 | 600805 | Epidermolysis bullosa, generalized atrophic benign | 226650 | AR |
| | | Epidermolysis bullosa, junctional, Herlitz type | 226700 | AR |
| | | Laryngoonychocutaneous syndrome | 245660 | AR |
| LAMB3 | 150310 | Amelogenesis imperfecta, type IA | 104530 | AD |
| | | Epidermolysis bullosa, junctional, Herlitz type | 226700 | AR |
| | | Epidermolysis bullosa, junctional, non-Herlitz type | 226650 | AR |
| LAMC2 | 150292 | Epidermolysis bullosa, junctional, Herlitz type | 226700 | AR |
| | | Epidermolysis bullosa, junctional, non-Herlitz type | 226650 | AR |
| LCA5 | 611408 | Leber congenital amaurosis 5 | 604537 | |
| LHCGR | 152790 | Leydig cell adenoma, somatic, with precocious puberty | 176410 | |
| | | Leydig cell hypoplasia with hypergonadotropic hypogonadism | 238320 | AR |
| | | Leydig cell hypoplasia with pseudohermaphroditism | 238320 | AR |
| | | Luteinizing hormone resistance, female | 238320 | AR |
| | | Precocious puberty, male | 176410 | AD |
| LHX3 | 600577 | Pituitary hormone deficiency, combined, 3 | 221750 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| LIFR | 151443 | Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome | 601559 | AR |
| LIPA | 613497 | Cholesteryl ester storage disease | 278000 | AR |
| | | Wolman disease | 278000 | AR |
| LIPH | 607365 | Hypotrichosis 7 | 604379 | AR |
| | | Woolly hair, autosomal recessive 2 with or without hypotrichosis | 604379 | AR |
| LOXHD1 | 613072 | Deafness, autosomal recessive 77 | 613079 | AR |
| LPL | 609708 | Combined hyperlipidemia, familial | 144250 | AD |
| | | Lipoprotein lipase deficiency | 238600 | AR |
| | | High density lipoprotein cholesterol level QTL 11 | | |
| LRPPRC | 607544 | Leigh syndrome, French-Canadian type | 220111 | AR |
| LYST | 606897 | Chediak-Higashi syndrome | 214500 | AR |
| MAN2B1 | 609458 | Mannosidosis, alpha-, types I and II | 248500 | AR |
| MCCC1 | 609010 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency | 210200 | AR |
| MCCC2 | 609014 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency | 210210 | AR |
| MCOLN1 | 605248 | Mucolipidosis IV | 252650 | AR |
| MECP2 | 300005 | Encephalopathy, neonatal severe | 300673 | XLR |
| | | Mental retardation, X-linked syndromic, Lubs type | 300260 | XLR |
| | | Mental retardation, X-linked, syndromic 13 | 300055 | XLR |
| | | Rett syndrome | 312750 | XLD |
| | | Rett syndrome, atypical | 312750 | XLD |
| | | Rett syndrome, preserved speech variant | 312750 | XLD |
| | | Autism susceptibility, X-linked 3 | 300496 | XL |
| MED17 | 603810 | Microcephaly, postnatal progressive, with seizures and brain atrophy | 613668 | AR |
| MEFV | 608107 | Familial Mediterranean fever, AD | 134610 | AD |
| | | Familial Mediterranean fever, AR | 249100 | AR |
| MFSD8 | 611124 | Ceroid lipofuscinosis, neuronal, 7 | 610951 | AR |
| | | Macular dystrophy with central cone involvement | 616170 | AR |
| MKS1 | 609883 | Bardet-Biedl syndrome 13 | 615990 | AR |
| | | Joubert syndrome 28 | 617121 | AR |
| | | Meckel syndrome 1 | 249000 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| MLC1 | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts | 604004 | AR |
| MMAA | 607481 | Methylmalonic aciduria, vitamin B12-responsive | 251100 | AR |
| MMAB | 607568 | Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type | 251110 | AR |
| MMACHC | 609831 | Methylmalonic aciduria and homocystinuria, cblC type | 277400 | AR |
| MMADHC | 611935 | Homocystinuria, cblD type, variant 1 | 277410 | AR |
| MPI | 154550 | Congenital disorder of glycosylation, type Ib | 602579 | AR |
| MPL | 159530 | Myelofibrosis with myeloid metaplasia, somatic | 254450 | |
| | | Thrombocythemia 2 | 601977 | SMu, AD |
| | | Thrombocytopenia, congenital amegakaryocytic | 604498 | AR |
| MPV17 | 137960 | Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) | 256810 | AR |
| MTHFR | 607093 | Homocystinuria due to MTHFR deficiency | 236250 | AR |
| | | Neural tube defects, susceptibility to | 601634 | AR |
| | | Schizophrenia, susceptibility to | 181500 | AD |
| | | Thromboembolism, susceptibility to | 188050 | AD |
| | | Vascular disease, susceptibility to | | |
| MTM1 | 300415 | Myotubular myopathy, X-linked | 310400 | XLR |
| MTTP | 157147 | Abetalipoproteinemia | 200100 | AR |
| | | Metabolic syndrome, protection against | 605552 | AD |
| MUSK | 601296 | Fetal akinesia deformation sequence | 208150 | AR |
| | | Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency | 616325 | AR |
| MUT | 609058 | Methylmalonic aciduria, mut(0) type | 251000 | AR |
| MYO7A | 276903 | Deafness, autosomal dominant 11 | 601317 | AD |
| | | Deafness, autosomal recessive 2 | 600060 | AR |
| | | Usher syndrome, type 1B | 276900 | AR |
| NAGLU | 609701 | Charcot-Marie-Tooth disease, axonal, type 2V | 616491 | AD |
| | | Mucopolysaccharidosis type IIIB (Sanfilippo B) | 252920 | AR |
| NAGS | 608300 | N-acetylglutamate synthase deficiency | 237310 | AR |
| NBN | 602667 | Aplastic anemia | 609135 | |
| | | Leukemia, acute lymphoblastic | 613065 | |
| | | Nijmegen breakage syndrome | 251260 | AR |
| NDUFS6 | 603848 | Mitochondrial complex I deficiency, nuclear type 9 | 618232 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| NEB | 161650 | Nemaline myopathy 2, autosomal recessive | 256030 | AR |
| NPC1 | 607623 | Niemann-Pick disease, type C1 | 257220 | AR |
| | | Niemann-Pick disease, type D | 257220 | AR |
| NPC2 | 601015 | Niemann-pick disease, type C2 | 607625 | AR |
| NPHP1 | 607100 | Joubert syndrome 4 | 609583 | AR |
| | | Nephronophthisis 1, juvenile | 256100 | AR |
| | | Senior-Loken syndrome-1 | 266900 | AR |
| NPHS1 | 602716 | Nephrotic syndrome, type 1 | 256300 | AR |
| NPHS2 | 604766 | Nephrotic syndrome, type 2 | 600995 | AR |
| NR2E3 | 604485 | Enhanced S-cone syndrome | 268100 | AR |
| | | Retinitis pigmentosa 37 | 611131 | AD, AR |
| NTRK1 | 191315 | Insensitivity to pain, congenital, with anhidrosis | 256800 | AR |
| | | Medullary thyroid carcinoma, familial | 155240 | AD |
| OCRL | 300535 | Dent disease 2 | 300555 | XLR |
| | | Lowe syndrome | 309000 | XLR |
| OPA3 | 606580 | 3-methylglutaconic aciduria, type III | 258501 | AR |
| | | Optic atrophy 3 with cataract | 165300 | AD |
| OTC | 300461 | Ornithine transcarbamylase deficiency | 311250 | XLR |
| PAH | 612349 | Phenylketonuria | 261600 | AR |
| | | Hyperphenylalaninemia, non-PKU mild | 261600 | AR |
| PANK2 | 606157 | HARP syndrome | 607236 | AR |
| | | Neurodegeneration with brain iron accumulation 1 | 234200 | AR |
| PC | 608786 | Pyruvate carboxylase deficiency | 266150 | AR |
| PCCA | 232000 | Propionicacidemia | 606054 | AR |
| PCCB | 232050 | Propionicacidemia | 606054 | AR |
| PCDH15 | 605514 | Deafness, autosomal recessive 23 | 609533 | AR |
| | | Usher syndrome, type 1D/F digenic | 601067 | DR, AR |
| | | Usher syndrome, type 1F | 602083 | AR |
| PDHA1 | 300502 | Pyruvate dehydrogenase E1-alpha deficiency | 312170 | XLD |
| PDHB | 179060 | Pyruvate dehydrogenase E1-beta deficiency | 614111 | |
| PEX1 | 602136 | Heimler syndrome 1 | 234580 | AR |
| | | Peroxisome biogenesis disorder 1A (Zellweger) | 214100 | AR |
| | | Peroxisome biogenesis disorder 1B (NALD/IRD) | 601539 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| PEX10 | 602859 | Peroxisome biogenesis disorder 6A (Zellweger) | 614870 | AR |
| | | Peroxisome biogenesis disorder 6B | 614871 | AR |
| PEX12 | 601758 | Peroxisome biogenesis disorder 3A (Zellweger) | 614859 | AR |
| | | Peroxisome biogenesis disorder 3B | 266510 | AR |
| PEX2 | 170993 | Peroxisome biogenesis disorder 5A (Zellweger) | 614866 | AR |
| | | Peroxisome biogenesis disorder 5B | 614867 | AR |
| PEX26 | 608666 | Peroxisome biogenesis disorder 7A (Zellweger) | 614872 | AR |
| | | Peroxisome biogenesis disorder 7B | 614873 | AR |
| PEX6 | 601498 | Heimler syndrome 2 | 616617 | AR |
| | | Peroxisome biogenesis disorder 4A (Zellweger) | 614862 | AR |
| | | Peroxisome biogenesis disorder 4B | 614863 | AD, AR |
| PEX7 | 601757 | Peroxisome biogenesis disorder 9B | 614879 | AR |
| | | Rhizomelic chondrodysplasia punctata, type 1 | 215100 | AR |
| PFKM | 610681 | Glycogen storage disease VII | 232800 | AR |
| PHGDH | 606879 | Neu-Laxova syndrome 1 | 256520 | AR |
| | | Phosphoglycerate dehydrogenase deficiency | 601815 | AR |
| PKHD1 | 606702 | Polycystic kidney disease 4, with or without hepatic disease | 263200 | AR |
| PLA2G6 | 603604 | Infantile neuroaxonal dystrophy 1 | 256600 | AR |
| | | Neurodegeneration with brain iron accumulation 2B | 610217 | AR |
| | | Parkinson disease 14, autosomal recessive | 612953 | AR |
| PMM2 | 601785 | Congenital disorder of glycosylation, type Ia | 212065 | AR |
| POLG | 174763 | Mitochondrial DNA depletion syndrome 4A (Alpers type) | 203700 | AR |
| | | Mitochondrial DNA depletion syndrome 4B (MNGIE type) | 613662 | AR |
| | | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) | 607459 | AR |
| | | Progressive external ophthalmoplegia, autosomal dominant 1 | 157640 | AD |
| | | Progressive external ophthalmoplegia, autosomal recessive 1 | 258450 | AR |
| POMGNT1 | 606822 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3 | 253280 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3 | 613151 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3 | 613157 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Retinitis pigmentosa 76 | 617123 | AR |
| POMT1 | 607423 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 | 236670 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1 | 613155 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1 | 609308 | AR |
| POMT2 | 607439 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 | 613150 | AR |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2 | 613156 | AR |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2 | 613158 | AR |
| PPT1 | 600722 | Ceroid lipofuscinosis, neuronal, 1 | 256730 | AR |
| PROP1 | 601538 | Pituitary hormone deficiency, combined, 2 | 262600 | AR |
| PRPS1 | 311850 | Arts syndrome | 301835 | XLR |
| | | Charcot-Marie-Tooth disease, X-linked recessive, 5 | 311070 | XLR |
| | | Deafness, X-linked 1 | 304500 | XL |
| | | Gout, PRPS-related | 300661 | XLR |
| | | Phosphoribosylpyrophosphate synthetase superactivity | 300661 | XLR |
| PSAP | 176801 | Combined SAP deficiency | 611721 | AR |
| | | Gaucher disease, atypical | 610539 | |
| | | Krabbe disease, atypical | 611722 | AR |
| | | Metachromatic leukodystrophy due to SAP-b deficiency | 249900 | AR |
| PTS | 612719 | Hyperphenylalaninemia, BH4-deficient, A | 261640 | AR |
| PUS1 | 608109 | Myopathy, lactic acidosis, and sideroblastic anemia 1 | 600462 | AR |
| PYGM | 608455 | McArdle disease | 232600 | AR |
| RAB23 | 606144 | Carpenter syndrome | 201000 | AR |
| RAG1 | 179615 | Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity | 609889 | |
| | | Combined cellular and humoral immune defects with granulomas | 233650 | AR |
| | | Omenn syndrome | 603554 | AR |
| | | Severe combined immunodeficiency, B cell-negative | 601457 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| RAG2 | 179616 | Combined cellular and humoral immune defects with granulomas | 233650 | AR |
| | | Omenn syndrome | 603554 | AR |
| | | Severe combined immunodeficiency, B cell-negative | 601457 | AR |
| RAPSN | 601592 | Fetal akinesia deformation sequence | 208150 | AR |
| | | Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency | 616326 | AR |
| RARS2 | 611524 | Pontocerebellar hypoplasia, type 6 | 611523 | AR |
| RDH12 | 608830 | Leber congenital amaurosis 13 | 612712 | AD, AR |
| RMRP | 157660 | Anauxetic dysplasia 1 | 607095 | AR |
| | | Cartilage-hair hypoplasia | 250250 | AR |
| | | Metaphyseal dysplasia without hypotrichosis | 250460 | AR |
| RPE65 | 180069 | Leber congenital amaurosis 2 | 204100 | AR |
| | | Retinitis pigmentosa 20 | 613794 | AR |
| RPGRIP1L | 610937 | COACH syndrome | 216360 | AR |
| | | Joubert syndrome 7 | 611560 | AR |
| | | Meckel syndrome 5 | 611561 | AR |
| RS1 | 300839 | Retinoschisis | 312700 | XLR |
| SACS | 604490 | Spastic ataxia, Charlevoix-Saguenay type | 270550 | AR |
| SAMHD1 | 606754 | Chilblain lupus 2 | 614415 | AD |
| | | Aicardi-Goutieres syndrome 5 | 612952 | AR |
| SBDS | 607444 | Shwachman-Diamond syndrome | 260400 | AR |
| | | Aplastic anemia, susceptibility to | 609135 | |
| SEPSECS | 613009 | Pontocerebellar hypoplasia type 2D | 613811 | AR |
| SERPINA1 | 107400 | Emphysema due to AAT deficiency | 613490 | AR |
| | | Emphysema-cirrhosis, due to AAT deficiency | 613490 | AR |
| | | Hemorrhagic diathesis due to antithrombin Pittsburgh | 613490 | AR |
| | | Pulmonary disease, chronic obstructive, susceptibility to | 606963 | |
| SGCA | 600119 | Muscular dystrophy, limb-girdle, autosomal recessive 3 | 608099 | AR |
| SGCB | 600900 | Muscular dystrophy, limb-girdle, autosomal recessive 4 | 604286 | AR |
| SGCG | 608896 | Muscular dystrophy, limb-girdle, autosomal recessive 5 | 253700 | AR |
| SGSH | 605270 | Mucopolysaccharidosis type IIIA (Sanfilippo A) | 252900 | AR |
| SLC12A3 | 600968 | Gitelman syndrome | 263800 | AR |
| SLC12A6 | 604878 | Agenesis of the corpus callosum with peripheral neuropathy | 218000 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| SLC17A5 | 604322 | Salla disease | 604369 | AR |
| | | Sialic acid storage disorder, infantile | 269920 | AR |
| SLC22A5 | 603377 | Carnitine deficiency, systemic primary | 212140 | AR |
| SLC25A13 | 603859 | Citrullinemia, adult-onset type II | 603471 | AR |
| | | Citrullinemia, type II, neonatal-onset | 605814 | AR |
| SLC25A15 | 603861 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome | 238970 | AR |
| SLC25A20 | 613698 | Carnitine-acylcarnitine translocase deficiency | 212138 | AR |
| SLC26A2 | 606718 | Achondrogenesis Ib | 600972 | AR |
| | | Atelosteogenesis, type II | 256050 | AR |
| | | De la Chapelle dysplasia | 256050 | AR |
| | | Diastrophic dysplasia | 222600 | AR |
| | | Diastrophic dysplasia, broad bone-platyspondylic variant | 222600 | AR |
| | | Epiphyseal dysplasia, multiple, 4 | 226900 | AR |
| SLC26A4 | 605646 | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct | 600791 | AR |
| | | Pendred syndrome | 274600 | AR |
| SLC37A4 | 602671 | Glycogen storage disease Ib | 232220 | AR |
| | | Glycogen storage disease Ic | 232240 | AR |
| SLC39A4 | 607059 | Acrodermatitis enteropathica | 201100 | AR |
| SLC4A11 | 610206 | Corneal dystrophy, Fuchs endothelial, 4 | 613268 | |
| | | Corneal endothelial dystrophy and perceptive deafness | 217400 | AR |
| | | Corneal endothelial dystrophy, autosomal recessive | 217700 | AR |
| SLC6A8 | 300036 | Cerebral creatine deficiency syndrome 1 | 300352 | XLR |
| SMN1 | 600354 | Spinal muscular atrophy-1 | 253300 | AR |
| | | Spinal muscular atrophy-2 | 253550 | AR |
| | | Spinal muscular atrophy-3 | 253400 | AR |
| | | Spinal muscular atrophy-4 | 271150 | AR |
| SMPD1 | 607608 | Niemann-Pick disease, type A | 257200 | AR |
| STAR | 600617 | Lipoid adrenal hyperplasia | 201710 | AR |
| SUMF1 | 607939 | Multiple sulfatase deficiency | 272200 | AR |
| TAT | 613018 | Tyrosinemia, type II | 276600 | AR |
| TCIRG1 | 604592 | Osteopetrosis, autosomal recessive 1 | 259700 | AR |
| TFR2 | 604720 | Hemochromatosis, type 3 | 604250 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| TGM1 | 190195 | Ichthyosis, congenital, autosomal recessive 1 | 242300 | AR |
| TH | 191290 | Segawa syndrome, recessive | 605407 | AR |
| TMEM216 | 613277 | Joubert syndrome 2 | 608091 | AR |
| | | Meckel syndrome 2 | 603194 | AR |
| TPP1 | 607998 | Ceroid lipofuscinosis, neuronal, 2 | 204500 | AR |
| | | Spinocerebellar ataxia, autosomal recessive 7 | 609270 | AR |
| TREX1 | 606609 | Aicardi-Goutieres syndrome 1, dominant and recessive | 225750 | AD, AR |
| | | Chilblain lupus | 610448 | AD |
| | | Vasculopathy, retinal, with cerebral leukodystrophy | 192315 | AD |
| | | Systemic lupus erythematosus, susceptibility to | 152700 | AD |
| TRIM37 | 605073 | Mulibrey nanism | 253250 | AR |
| TSEN2 | 608753 | Pontocerebellar hypoplasia type 2B | 612389 | AR |
| TSEN34 | 608754 | Pontocerebellar hypoplasia type 2C | 612390 | |
| TSEN54 | 608755 | Pontocerebellar hypoplasia type 5 | 610204 | AR |
| | | Pontocerebellar hypoplasia type 2A | 277470 | AR |
| | | Pontocerebellar hypoplasia type 4 | 225753 | AR |
| TSFM | 604723 | Combined oxidative phosphorylation deficiency 3 | 610505 | AR |
| TSHB | 188540 | Hypothyroidism, congenital, nongoitrous 4 | 275100 | AR |
| TTC37 | 614589 | Trichohepatoenteric syndrome 1 | 222470 | AR |
| TTPA | 600415 | Ataxia with isolated vitamin E deficiency | 277460 | AR |
| TYMP | 131222 | Mitochondrial DNA depletion syndrome 1 (MNGIE type) | 603041 | AR |
| TYR | 606933 | Albinism, oculocutaneous, type IA | 203100 | AR |
| | | Albinism, oculocutaneous, type IB | 606952 | |
| | | Waardenburg syndrome/albinism, digenic | 103470 | AD |
| | | Skin/hair/eye pigmentation 3, blue/green eyes | 601800 | |
| | | Skin/hair/eye pigmentation 3, light/dark/freckling skin | 601800 | |
| | | Melanoma, cutaneous malignant, susceptibility to, 8 | 601800 | |
| UGT1A1 | 191740 | Crigler-Najjar syndrome, type I | 218800 | AR |
| | | Crigler-Najjar syndrome, type II | 606785 | AR |
| | | Hyperbilirubinemia, familial transient neonatal | 237900 | AR |
| | | Bilirubin, serum level of, QTL1 | 601816 | |
| | | Gilbert syndrome | 143500 | AR |
| USH1C | 605242 | Deafness, autosomal recessive 18A | 602092 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

| | | | | |
|---|---|---|---|---|
| | | Usher syndrome, type 1C | 276904 | AR |
| USH2A | 608400 | Retinitis pigmentosa 39 | 613809 | |
| | | Usher syndrome, type 2A | 276901 | AR |
| VPS13A | 605978 | Choreoacanthocytosis | 200150 | AR |
| VPS13B | 607817 | Cohen syndrome | 216550 | AR |
| VRK1 | 602168 | Pontocerebellar hypoplasia type 1A | 607596 | AR |
| WAS | 300392 | Neutropenia, severe congenital, X-linked | 300299 | XLR |
| | | Thrombocytopenia, X-linked | 313900 | XLR |
| | | Thrombocytopenia, X-linked, intermittent | 313900 | XLR |
| | | Wiskott-Aldrich syndrome | 301000 | XLR |
| WNT10A | 606268 | Odontoonychodermal dysplasia | 257980 | AR |
| | | Schopf-Schulz-Passarge syndrome | 224750 | AR |
| | | Tooth agenesis, selective, 4 | 150400 | AD, AR |
| XPA | 611153 | Xeroderma pigmentosum, group A | 278700 | AR |
| XPC | 613208 | Xeroderma pigmentosum, group C | 278720 | AR |
| ZFYVE26 | 612012 | Spastic paraplegia 15, autosomal recessive | 270700 | AR |



Legal Head Office
00138 Roma - Via di Castel Giubileo, 11
C.F. e P. Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Lab and Medical Practice Rome
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 (12 linee PBX) - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Lab and Medical Practice Milan
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it