

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

Gene	OMIM Gene	Disease	OMIM Disease	Inheritance
ACADM	<a href="#">607008</a>	Acyl-CoA dehydrogenase, medium chain, deficiency of	<a href="#">201450</a>	AR
AGXT	<a href="#">604285</a>	Hyperoxaluria, primary, type 1	<a href="#">259900</a>	AR
ARSA	<a href="#">607574</a>	Metachromatic leukodystrophy	<a href="#">250100</a>	AR
ATP7B	<a href="#">606882</a>	Wilson disease	<a href="#">277900</a>	AR
BTD	<a href="#">609019</a>	Biotinidase deficiency	<a href="#">253260</a>	AR
CBS	<a href="#">613381</a>	Homocystinuria	<a href="#">236200</a>	AR
CFTR	<a href="#">602421</a>	Cystic fibrosis	<a href="#">219700</a>	AR
DHCR7	<a href="#">602858</a>	Smith-Lemli-Opitz syndrome	<a href="#">270400</a>	AR
EMD	<a href="#">300384</a>	Emery-Dreifuss muscular dystrophy 1, X-linked	<a href="#">310300</a>	XLR
FMR1	<a href="#">309550</a>	Fragile X syndrome	<a href="#">300624</a>	XLD
GAA	<a href="#">606800</a>	Glycogen storage disease II	<a href="#">232300</a>	AR
GALC	<a href="#">606890</a>	Krabbe disease	<a href="#">245200</a>	AR
GALT	<a href="#">606999</a>	Galactosemia	<a href="#">230400</a>	AR
GBA	<a href="#">606463</a>	Gaucher disease type I-II-III-IIIc-Perinatal Lethal	<a href="#">608013</a> <a href="#">230800</a> <a href="#">230900</a> <a href="#">231000</a> <a href="#">231005</a>	AR
GJB1	<a href="#">304040</a>	Charcot-Marie-Tooth neuropathy, X-linked	<a href="#">302800</a>	XLD
GJB2	<a href="#">121011</a>	Deafness, autosomal recessive 1A	<a href="#">220290</a>	AR
GJB6	<a href="#">604418</a>	Deafness, autosomal recessive 1B	<a href="#">612645</a>	AR
GLA	<a href="#">300644</a>	Fabry disease	<a href="#">301500</a>	XL
HADHA	<a href="#">600890</a>	LCHAD deficiency	<a href="#">609016</a>	AR
HBA1	<a href="#">141800</a>	Alpha-Thalassemia	<a href="#">604131</a>	AR
HBA2	<a href="#">141850</a>	Alpha-Thalassemia	<a href="#">604131</a>	AR
HBB	<a href="#">141900</a>	Beta-Thalassemia, Sickle cell anemia	<a href="#">603903</a> <a href="#">613985</a>	AR
HEXA	<a href="#">606869</a>	Tay-Sachs disease	<a href="#">272800</a>	AR
MEFV	<a href="#">608107</a>	Familial Mediterranean fever	<a href="#">249100</a>	AR
MMACHC	<a href="#">609831</a>	Methylmalonic aciduria and homocystinuria, cblC type	<a href="#">277400</a>	AR
PAH	<a href="#">612349</a>	Phenylketonuria	<a href="#">261600</a>	AR

**ROMA**

Laboratori e Studi Medici  
Sede Legale e Laboratorio  
di Ricerca e Sviluppo  
in Genetica Molecolare  
Via Castel Giubileo, 11  
00138

Laboratorio Genetica  
Medica e Diagnostica  
Molecolare  
Prelevi e Consulenze  
Via Castel Giubileo, 62  
00138

**MILANO**

Laboratorio Genetica  
Molecolare e Studi Medici  
Via Enrico Cialdini, 16  
(Affori Centre)  
20161

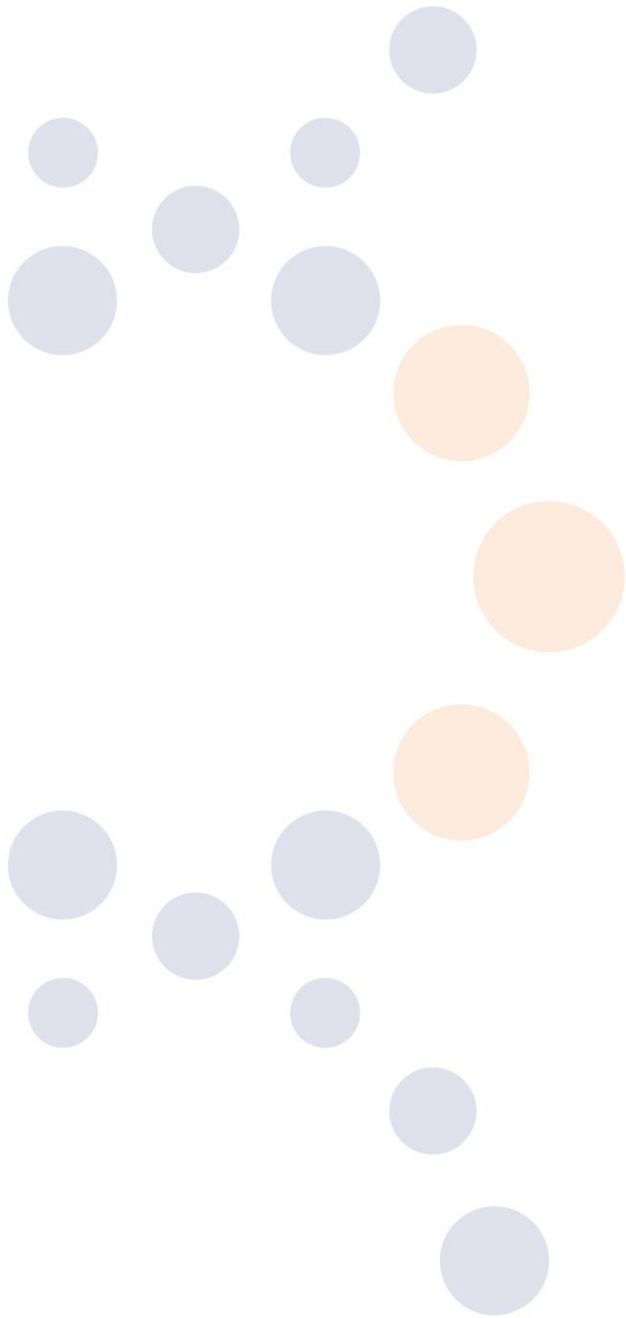
**FIRENZE**

Laboratorio e studi medici  
Via Cavour, 168r  
50121



COMPANY WITH  
QUALITY SYSTEM  
CERTIFIED BY DNV  
ISO 9001

PMM2	<u>601785</u>	Congenital disorder of glycosylation, type Ia	<u>212065</u>	AR
SERPINA1	<u>107400</u>	Alpha-1-Antitrypsin Deficiency	<u>613490</u>	AR
SLC26A2	<u>606718</u>	Achondrogenesis Ib	<u>600972</u>	AR
SMN1	<u>600354</u>	Spinal muscular atrophy (SMA1)	<u>253300</u>	AR



**ROMA**

**Laboratori e Studi Medici**  
Sede Legale e Laboratorio  
di Ricerca e Sviluppo  
in Genetica Molecolare  
Via Castel Giubileo, 11  
00138

**Laboratorio Genetica  
Medica e Diagnostica  
Molecolare**  
Prelievi e Consulenze  
Via Castel Giubileo, 62  
00138

**MILANO**

**Laboratorio Genetica  
Molecolare e Studi Medici**  
Via Enrico Cialdini, 16  
(Affori Centre)  
20161

**FIRENZE**

**Laboratorio e studi medici**  
Via Cavour, 168r  
50121

