FETALDNA
IL TEST PRENATALE DI ULTIMA GENERAZIONE

ALTAMEDICA
ARTEMISIA

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

Partner femminile	**<nome>**	Data di Nascita	**<datanasc>**
Identificativo del partner femminile	**<pid>**		
Tipo di Gravidanza	**Spontanea**	Gestazione	**Singola**
Partner maschile		Data di Nascita	**<datanasc>**
Identificativo del partner maschile	**<pid>P**		
Accettazione N°	**<NrAcc>**	Data Refertazione	**<timestamp>**

RISULTATI

Sesso Fetale | Frazione Fetale **%**

FETALDNA - INDAGINI SUL FETO	ESITO
Principali aneuploidie cromosomiche fetali	
Cromosoma 13 (sindrome di Patau)	**NEGATIVO**
Cromosoma 18 (sindrome di Edwards)	**NEGATIVO**
Cromosoma 21 (sindrome di Down)	**NEGATIVO**
Aneuploidie di tutti i cromosomi fetali (1-22)	**NEGATIVO**
Principali aneuploidie dei cromosomi sessuali: X0, XXY, XXX, XYY	**NEGATIVO**
Sindromi da Microdelezioni Sindrome di Wolf-Hirschhorn, Sindrome HNPP, Sindrome di Jacobsen, Sindrome da delezione 18q, Sindrome da delezione 1p36, Sindrome di Alagille, Sindrome di Angelman, Sindrome di Rubinstein-Taybi, Sindrome di DiGeorge, Sindrome di WAGR, Sindrome di Cri-du-chat, Sindrome di Potocki-Shaffer, Sindrome di Langer-Giedion, Sindrome di Miller-Dieker, Sindrome di Smith-Magenis, Sindrome da delezione 1q21.1, Sindrome di Prader-Willi, Sindrome di Kleefstra, Sindrome di Williams, Sindrome di Phelan-Mcdermid, Sindrome di Koolen-de-Vries	**NEGATIVO**
Mutazioni Malattie Monogeniche Fetali Sindrome di Apert, Sindrome di Crouzon, Sindrome di Pfeiffer, Sindrome di Leopard, Sindrome di Noonan, Acondroplasia, Ipocondroplasia, Displasia tanatofora.	**NEGATIVO**

CARRIER SCREENING - INDAGINI SULLA COPPIA	ESITO
Analisi geni come da elenco per ulteriori dettagli visualizzare l'elenco completo delle patologie	Non sono presenti in entrambi i genitori mutazioni, analoghe o diverse, sullo stesso gene. Oppure in caso di positività: sono presenti , a livello del gene….., le seguenti mutazioni on è presente la stessa mutazione (contemporaneamente) in entrambi i genitori. **Si consiglia diagnosi prenatale mirata sul feto**

Dott. Claudio Dello Russo

Controllo qualità

Prof. Dott. Alvaro Mesoraca
Specialista in Genetica Medica

Il Direttore

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NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
1.AAAS	Achalasia-addisonianism-alacrimia syndrome		
2.ABCA12	Ichthyosis, autosomal recessive 4B (harlequin)		
	Ichthyosis, congenital, autosomal recessive 4A		
	Surfactant metabolism dysfunction, pulmonary, 3		
3.ABCA4	Cone-rod dystrophy 3		
	Fundus flavimaculatus		
	Macular degeneration, age-related, 2		
	Retinal dystrophy, early-onset severe		
	Retinitis pigmentosa 19		
	Stargardt disease 1		
4.ABCB11	Cholestasis, benign recurrent intrahepatic, 2		
	Cholestasis, progressive familial intrahepatic 2		
5.ABCB4	Cholestasis, intrahepatic, of pregnancy, 3		
	Cholestasis, progressive familial intrahepatic 3		
	Gallbladder disease 1		
6.ABCB7	Anemia, sideroblastic, with ataxia		
7.ABCC2	Dubin-Johnson syndrome		
8.ABCC6	Arterial calcification, generalized, of infancy, 2		
	Pseudoxanthoma elasticum		
	Pseudoxanthoma elasticum, forme fruste		
9.ABCC8	Diabetes mellitus, noninsulin-dependent		
	Diabetes mellitus, permanent neonatal		
	Diabetes mellitus, transient neonatal 2		
	Hyperinsulinemic hypoglycemia, familial, 1		
	Hypoglycemia of infancy, leucine-sensitive		
10.ABCD1	Adrenoleukodystrophy		
11.ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type		
12.ACAD8	Isobutyryl-CoA dehydrogenase deficiency		
13.ACAD9	Acyl-CoA dehydrogenase 9 deficiency		
14.ACADL	ACYL-CoA DEHYDROGENASE, LONG-CHAIN		
15.ACADM	Medium chain acyl-CoA dehydrogenase deficiency		
16.ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of		
17.ACADSB	2-methylbutyrylglycinuria		
18.ACADVL	Very long chain acyl-CoA dehydrogenase deficiency		
19.ACAT1	Ketoacidosis due to beta-ketothiolase deficiency		
20.ACE	Angiotensin I-converting enzyme, benign serum increase		
	Microvascular complications of diabetes 3		
	Myocardial infarction, susceptibility to		
	Renal tubular dysgenesis		
	SARS, progression of		
	Stroke, hemorrhagic		
21.ACOX1	Peroxisomal acyl-CoA oxidase deficiency		
22.ACSF3	Combined malonic and methylmalonic aciduria		
23.ACSL4	Mental retardation, X-linked 63		
24.ACTN4	Glomerulosclerosis, focal segmental, 1		
25.ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency		
26.ADAMTS 13	Thrombotic thrombocytopenic purpura, familial		

GENE	MALATTIA	<pid>	<pid>P
27.ADAMTS 2	Ehlers-Danlos syndrome, dermatosparaxis type		
28.ADAMTS L2	Geleophysic dysplasia 1		
29.ADCK3	Autosomal recessive ataxia due to ubiquinone deficiency		
30.ADK	Hypermethioninemia due to adenosine kinase deficiency		
31.AFF2	Mental retardation, X-linked, FRAXE type		
32.AGA	Aspartylglucosaminuria		
33.AGL	Glycogen storage disease due to glycogen debranching enzyme deficiency		
34.AGPS	Rhizomelic chondrodysplasia punctata type 3		
35.AGT	Hypertension, essential, susceptibility to		
	Preeclampsia, susceptibility to		
	Renal tubular dysgenesis		
36.AGTR1	Hypertension, essential		
	Renal tubular dysgenesis		
37.AGTR2	ANGIOTENSIN II RECEPTOR, TYPE 2		
38.AGXT	Hyperoxaluria, primary, type 1		
39.AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase		
40.AHI1	Joubert syndrome with ocular defect		
41.AIPL1	Cone-rod dystrophy		
	Leber congenital amaurosis 4		
	Retinitis pigmentosa, juvenile		
42.AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia		
43.ALAS2	Anemia, sideroblastic, 1		
	Protoporphyria, erythropoietic, X-linked		
44.ALDH3A2	Sjogren-Larsson syndrome		
45.ALDH4A1	Hyperprolinemia, type II		
46.ALDH5A1	4-hydroxybutyric aciduria		
47.ALDH7A1	Epilepsy, pyridoxine-dependent		
48.ALDOA	Glycogen storage disease XII		
49.ALDOB	Hereditary fructose intolerance		
50.ALG1	Congenital disorder of glycosylation type Ik		
51.ALG12	Congenital disorder of glycosylation, type Ig		
52.ALG2	Congenital disorder of glycosylation, type Ii		
53.ALG3	Congenital disorder of glycosylation, type Id		
54.ALG6	Congenital disorder of glycosylation type Ic		
55.ALG8	Congenital disorder of glycosylation, type Ih		
56.ALG9	Congenital disorder of glycosylation, type II		
57.ALMS1	Alström syndrome		
58.ALPL	Childhood-onset hypophosphatasia		
	Infantile hypophosphatasia		
59.ALS2	Amyotrophic lateral sclerosis 2, juvenile		
	Primary lateral sclerosis, juvenile		
	Spastic paralysis, infantile onset ascending		
60.AMACR	Alpha-methylacyl-Coa Racemase deficiency		
	Congenital bile acid synthesis defect type 4		
61.AMPD1	Myopathy due to myoadenylate deaminase deficiency		
62.AMT	Glycine encephalopathy		

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COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
63.ANO5	Gnathodiaphyseal dysplasia		
	Miyoshi muscular dystrophy 3		
	Muscular dystrophy, limb-girdle, autosomal recessive		
64.ANTXR2	Hyaline fibromatosis syndrome		
65.AP1S1	MEDNIK syndrome		
66.AP1S2	Mental retardation, X-linked syndromic 5		
67.AP3B1	Hermansky-Pudlak syndrome 2		
68.APTX	Ataxia - oculomotor apraxia type 1		
69.AQP2	Diabetes insipidus, nephrogenic		
70.AR	Complete androgen insensitivity syndrome		
	Kennedy disease		
	Partial androgen insensitivity syndrome		
71.ARG1	Argininemia		
72.ARHGEF6	Mental retardation, X-linked 46		
73.ARHGEF9	Epileptic encephalopathy, early infantile, 8		
74.ARL13B	Joubert syndrome 8		
75.ARL6	Bardet-Biedl syndrome 1, modifier of		
	Bardet-Biedl syndrome 3		
	Retinitis pigmentosa 55		
76.ARSA	Metachromatic leukodystrophy		
77.ARSB	Mucopolysaccharidosis type 6		
78.ARSE	Brachytelephalangic chondrodysplasia punctata		
79.ARSF	ARYLSULFATASE F		
80.ARX	Early infantile epileptic encephalopathy		
81.ASL	Argininosuccinic aciduria		
82.ASNS	Asparagine synthetase deficiency		
83.ASPA	Canavan disease		
84.ASPM	Microcephaly 5, primary, autosomal recessive		
85.ASS1	Citrullinemia type I		
86.ATIC	AICA-ribosiduria due to ATIC deficiency		
87.ATM	Ataxia-telangiectasia		
88.ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type		
	Parkinsonism with spasticity, X-linked		
89.ATP6V0A 2	Cutis laxa, autosomal recessive, type IIA		
	Wrinkly skin syndrome		
90.ATP6V1B 1	Renal tubular acidosis with deafness		
91.ATP7A	Menkes disease		
	Occipital horn syndrome		
	X-linked distal spinal muscular atrophy		
92.ATP7B	Wilson disease		
93.ATP8B1	Cholestasis, benign recurrent intrahepatic		
	Cholestasis, intrahepatic, of pregnancy, 1		
	Cholestasis, progressive familial intrahepatic 1		
94.ATR	Seckel syndrome		
95.ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic		
	Alpha-thalassemia/mental retardation syndrome		
	Mental retardation-hypotonic facies syndrome, X-linked		

GENE	MALATTIA	<pid>	<pid>P
96.AUH	3-methylglutaconic aciduria type 1		
97.B4GALT1	Congenital disorder of glycosylation type 2d		
98.B9D2	Joubert syndrome 34		
	Meckel syndrome 10		
99.BBS1	Bardet-Biedl syndrome 1		
100.BBS10	Bardet-Biedl syndrome 10		
101.BBS12	Bardet-Biedl syndrome 12		
102.BBS2	Bardet-Biedl syndrome 2Z		
	Retinitis pigmentosa 74		
103.BCHE	Apnea, postanesthetic, susceptibility to, due to BCHE deficiency		
	Butyrylcholinesterase deficiency		
104.BCKDHA	Maple syrup urine disease (gene BCKDHA)		
105.BCKDHB	Maple syrup urine disease (gene BCKDHB)		
106.BCOR	Microphthalmia, syndromic 2		
107.BCS1L	Björnstad syndrome		
	GRACILE syndrome		
	Isolated CoQ-cytochrome C reductase deficiency		
	Leigh syndrome		
108.BEST1	Bestrophinopathy, autosomal recessive		
	Macular dystrophy, vitelliform, 2		
	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma		
	Retinitis pigmentosa, concentric		
	Retinitis pigmentosa-50		
	Vitreoretinochoroidopathy		
109.BLM	Bloom syndrome		
110.BRCA2	Breast cancer, male, susceptibility to		
	Breast-ovarian cancer, familial, 2		
	Fanconi anemia, complementation group D1		
	Glioblastoma 3		
	Medulloblastoma		
	Pancreatic cancer 2		
	Prostate cancer		
	Wilms tumor		
111.BRIP1	Breast cancer, early-onset, susceptibility to		
	Fanconi anemia, complementation group J		
	Myasthenic syndrome, fast-channel congenital		
	Myasthenic syndrome, slow-channel congenital		
112.BRWD3	Mental retardation, X-linked 93		
113.BSCL2	Encephalopathy, progressive, with or without lipodystrophy		
	Lipodystrophy, congenital generalized, type 2		
	Neuropathy, distal hereditary motor, type VA		
	Silver spastic paraplegia syndrome		
114.BSND	Bartter syndrome, type 4a		
	Sensorineural deafness with mild renal dysfunction		
115.BTD	Biotinidase deficiency		
116.BTK	Isolated growth hormone deficiency type III		
	X-linked agammaglobulinemia		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
117.C10orf2	Infantile onset spinocerebellar ataxia		
118..3	C3 deficiency		
	Hemolytic uremic syndrome, atypical, susceptibility to, 5		
	Macular degeneration, age-related, 9		
119.CA2	Osteopetrosis with renal tubular acidosis		
120.CANT1	Desbuquois dysplasia 1		
	Epiphyseal dysplasia, multiple, 7		
121.CAPN3	Muscular dystrophy, limb-girdle, autosomal dominant 4		
	Muscular dystrophy, limb-girdle, autosomal recessive 1		
122.CASK	FG syndrome 4		
	Mental retardation and microcephaly with pontine and cerebellar hypoplasia		
	Mental retardation, with or without nystagmus		
123.CASP10	Autoimmune lymphoproliferative syndrome, type II		
124.CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2		
125.CBS	Classical homocystinuria		
126.CC2D2A	COACH syndrome		
	Joubert syndrome 9		
	Meckel syndrome 6		
127.CCDC103	Ciliary dyskinesia, primary, 17		
128.CCDC39	Ciliary dyskinesia, primary, 14		
129.CD19	Immunodeficiency, common variable, 3		
130.CD247	Immunodeficiency 25		
131.CD2AP	Glomerulosclerosis, focal segmental, 3		
132.CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect		
133.CD3D	Immunodeficiency 19		
134.CD3E	Immunodeficiency 18, SCID variant		
135.CD3G	Immunodeficiency 17, CD3 gamma deficient		
136.CD40LG	X-linked hyper-IgM syndrome		
137.CDH23	Autosomal recessive nonsyndromic sensorineural deafness type DFNB12		
138.CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy		
	Hypotrichosis, congenital, with juvenile macular dystrophy		
139.CDHR1	Cone-rod dystrophy 15		
	Retinitis pigmentosa 65		
140.CDK5RAP 2	Microcephaly 3, primary, autosomal recessive		
141.CDKL5	Epileptic encephalopathy, early infantile, 2		
142.CENPJ	Microcephaly 6, primary, autosomal recessive		
	Seckel syndrome 4		
143.CEP152	Microcephaly 9, primary, autosomal recessive		
	Seckel syndrome 5		
144.CEP290	Joubert syndrome with oculorenal defect 5		
	Senior-Loken syndrome		
145.CERKL	Retinitis pigmentosa 26		
146.CFH	Basal laminar drusen		
	Complement factor H deficiency		
	Hemolytic uremic syndrome, atypical, susceptibility to, 1		
	Macular degeneration, age-related, 4		
147.CFP	Properdin deficiency, X-linked		

GENE	MALATTIA	<pid>	<pid>P
148.CFTR	Cystic fibrosis; mucoviscidosis		
149.CHM	Choroideremia		
150.CHRNA1	Multiple pterygium syndrome, lethal type		
	Myasthenic syndrome, fast-channel congenital		
	Myasthenic syndrome, slow-channel congenital		
151.CHRND	Multiple pterygium syndrome, lethal type		
	Myasthenic syndrome, fast-channel congenital		
	Myasthenic syndrome, slow-channel congenital		
152.CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel		
	Myasthenic syndrome, congenital, 4B, fast-channel		
	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency		
153.CHRNG	Escobar syndrome		
	Multiple pterygium syndrome, lethal type		
	Myasthenia gravis, neonatal transient		
154.CHST6	Macular corneal dystrophy		
155.CIITA	Bare lymphocyte syndrome, type II, complementation group A		
	Rheumatoid arthritis, susceptibility to		
156.CLCN1	Myotonia congenita, dominant		
	Myotonia congenita, recessive		
	Myotonia levior, recessive		
157.CLCN5	Dent disease		
	Hypophosphatemic rickets		
	Nephrolithiasis, type I		
	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990		
158.CLCN7	Autosomal recessive malignant osteopetrosis 4		
159.CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis 607626		
160.CLDN14	Deafness, autosomal recessive 29		
161.CLDN19	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement		
162.CLN3	Juvenile neuronal ceroid lipofuscinosis 3		
163.CLN5	Late infantile neuronal ceroid lipofuscinosis 5		
164.CLN6	Adult neuronal ceroid lipofuscinosis 4A		
	Late infantile neuronal ceroid lipofuscinosis 6		
165.CLN8	Late infantile neuronal ceroid lipofuscinosis 8		
	Progressive epilepsy - intellectual deficit, Finnish type		
166.CLRN1	Usher syndrome type 3A		
167.CNGA1	Retinitis pigmentosa 49		
168.CNGA3	Achromatopsia 2		
169.CNGB1	Retinitis pigmentosa 45		
170.CNGB3	Achromatopsia 3		
	Macular degeneration, juvenile		
171.COG1	Congenital disorder of glycosylation, type IIg		
172.COG7	Congenital disorder of glycosylation, type IIe		
173.COG8	Congenital disorder of glycosylation, type IIh		
174. COL11A1	Fibrochondrogenesis 1		
	Lumbar disc herniation, susceptibility to		
	Marshall syndrome		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
175. OL17A1	Generalized junctional epidermolysis bullosa, non-Herlitz type		
176.COL18A1	Knobloch syndrome, type 1		
177.COL1A1	Caffey disease		
	Ehlers-Danlos syndrome, type I		
	Ehlers-Danlos syndrome, type VIIA		
	Osteogenesis imperfecta, type I		
	Osteogenesis imperfecta, type II		
	Osteogenesis imperfecta, type III		
	Osteogenesis imperfecta, type IV		
178.COL1A2	Ehlers-Danlos syndrome, cardiac valvular type		
179.COL2A1	Achondrogenesis, type II or hypochondrogenesis		
	Avascular necrosis of the femoral head		
	Czech dysplasia		
	Epiphyseal dysplasia, multiple, with myopia and deafness		
	Kniest dysplasia		
	Legg-Calve-Perthes disease		
	Osteoarthritis with mild chondrodysplasia		
	Platyspondylic skeletal dysplasia, Torrance type		
	SED congenita		
	SMED Strudwick type		
	Spondyloepiphyseal dysplasia, Stanescu type		
	Spondyloperipheral dysplasia		
	Stickler sydrome, type I, nonsyndromic ocular		
	Stickler syndrome, type I		
	Vitreoretinopathy with phalangeal epiphyseal dysplasia		
180.COL4A3	Alport syndrome autosomal recessive (gene COL4A3)		
181.COL4A4	Alport syndrome autosomal recessive (gene COL4A4)		
182.COL4A5	Alport syndrome		
183.COL6A1	Bethlem myopathy		
	Ullrich congenital muscular dystrophy		
184.COL6A2	Bethlem myopathy		
	Ullrich congenital muscular dystrophy		
185.COL6A3	Bethlem myopathy		
	Ullrich congenital muscular dystrophy		
186.COL7A1	Dystrophic epidermolysis bullosa pruriginosa		
	Severe generalized recessive dystrophic epidermolysis bullosa		
187.COL9A1	Epiphyseal dysplasia, multiple, 6		
	Stickler syndrome, type IV		
188.COL9A2	Epiphyseal dysplasia, multiple, 2		
	Stickler syndrome, type V		
189.COQ2	Leigh syndrome with nephrotic syndrome		
190.COQ9	Coenzyme Q10 deficiency, primary, 5		
191.COX10	Leigh syndrome due to mitochondrial COX4 deficiency		
	Mitochondrial complex IV deficiency		
192.COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2		
	Leigh syndrome due to cytochrome c oxidase deficiency		
193.COX6B1	Mitochondrial complex IV deficiency		

GENE	MALATTIA	<pid>	<pid>P
194.CPS1	Carbamoylphosphate synthetase deficiency		
195.CPT1A	Carnitine palmitoyl transferase 1A deficiency		
196.CPT2	Carnitine palmitoyl transferase II deficiency, infantile form		
	Carnitine palmitoyl transferase II deficiency, neonatal form		
197.CRB1	Leber congenital amaurosis 8		
	Pigmented paravenous chorioretinal atrophy		
	Retinitis pigmentosa-12		
198.CRLF1	Cold-induced sweating syndrome		
199.CRTAP	Osteogenesis imperfecta type VII		
200.CRX	Cone-rod retinal dystrophy-2		
	Leber congenital amaurosis 7		
201.CSTB	Unverricht-Lundborg disease		
202.CTH	Cystathioninuria		
	Homocysteine, total plasma, elevated		
203.CTNS	Cystinosis		
204.CTSC	Haim-Munk syndrome		
	Papillon-Lefevre syndrome		
	Periodontitis 1, juvenile		
205.CTSD	Adult neuronal ceroid lipofuscinosis 10		
206.CTSK	Pycnodysostosis		
207.CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type)		
208.CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA		
209.CYBB	Chronic granulomatous disease, X-linked		
	Immunodeficiency 34, mycobacteriosis, X-linked		
210.CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete		
211.CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency		
	Aldosteronism, glucocorticoid-remediable		
212.CYP11B2	Aldosterone to renin ratio raised		
	Hypoaldosteronism, congenital, due to CMO I deficiency		
	Hypoaldosteronism, congenital, due to CMO II deficiency		
	Low renin hypertension, susceptibility to		
213.CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency		
214.CYP19A1	Aromatase deficiency		
	Aromatase excess syndrome		
215.CYP1B1	Anterior segment dysgenesis 6, multiple subtypes		
	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset		
216.CYP27A1	Cerebrotendinous xanthomatosis		
217.CYP27B1	Vitamin D-dependent rickets, type I		
218.CYP4V2	Bietti crystalline corneoretinal dystrophy		
219.CYP7B1	Bile acid synthesis defect, congenital, 3		
	Spastic paraplegia 5A, autosomal recessive		
220.D2HGDH	D-2-hydroxyglutaric aciduria		
221.DBT	Classic maple syrup urine disease		
222.DCLRE1C	Omenn syndrome		
	Severe combined immunodeficiency due to DCLRE1C deficiency		
223.DCX	Lissencephaly, X-linked		
	Subcortical laminal heteropia, X-linked		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
224.DDB2	Xeroderma pigmentosum complementation group E		
225.DDC	Aromatic L-amino acid decarboxylase deficiency		
226.DFNB59	Deafness, autosomal recessive 59		
227.DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due toDGUOK deficiency 3		
228.DHCR24	Desmosterolosis		
229.DHCR7	Smith-Lemli-Opitz syndrome		
230.DHDDS	Congenital disorder of glycosylation, type 1bb		
	Developmental delay and seizures with or without movement abnormalities		
	Retinitis pigmentosa 59		
231.DKC1	Dyskeratosis congenita X-linked		
	Hoyeraal-Hreidarsson syndrome		
232.DLD	Leigh syndrome		
	Maple syrup urine disease		
233.DLG3	Mental retardation, X-linked 90		
234.DLL3	Autosomal recessive spondylocostal dysostosis 1		
235.DMD	Becker muscular dystrophy		
	Duchenne muscular dystrophy		
236.DMP1	Autosomal recessive hypophosphatemic rickets 1		
237.DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus		
238.DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus		
239.DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus		
240.DNAJC19	Dilated cardiomyopathy with ataxia		
241.DNAL1	Ciliary dyskinesia, primary, 16		
242.DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1		
243.DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive		
244.DOK7	Fetal akinesia deformation sequence		
	Myasthenia, limb-girdle, familial		
245.DOLK	Congenital disorder of glycosylation, type Im		
246.DPAGT1	Congenital disorder of glycosylation type 1j		
247.DPM1	Congenital disorder of glycosylation type 1e		
248.DPYD	Dihydropyrimidine dehydrogenase deficiency		
249.DSP	Lethal acantholytic epidermolysis bullosa		
250.DUOX2	Thyroid dyshormonogenesis 6		
251.DUOXA2	Thyroid dyshormonogenesis 5		
252.DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly		
253.DYSF	Miyoshi muscular dystrophy 1		
	Muscular dystrophy, limb-girdle, autosomal recessive 2		
	Myopathy, distal, with anterior tibial onset		
254.EDA	Muscular dystrophy, limb-girdle, autosomal recessive 2		
	Ectodermal dysplasia 1, hypohidrotic, X-linked		
	Tooth agenesis, selective, X-linked 1		
255.EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant		
	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive		
	Hair morphology 1, hair thickness		
256.EDN3	Waardenburg-Shah syndrome 4B		

GENE	MALATTIA	<pid>	<pid>P
257.EDNRB	ABCD syndrome		
	Waardenburg-Shah syndrome 4A		
258.EFEMP2	Cutis laxa, autosomal recessive, type IB		
259.EFNB1	Craniofrontonasal dysplasia		
260.EGR2	Charcot-Marie-Tooth disease type 4E		
261.EIF2AK3	Wolcott-Rallison syndrome		
262.EIF2B5	Leukoencephalopathy with vanishing white matter		
	Ovarioleukodystrophy		
263.ELK1	MEMBER OF ETS ONCOGENE FAMILY		
264.EMD	Emery-Dreifuss muscular dystrophy 1, X-linked		
265.ENO3	Glycogen storage disease XIII		
266.ENPP1	Autosomal recessive hypophosphatemic rickets 2		
267.EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)		
268.ERBB3	Lethal congenital contractural syndrome 2		
269.ERCC2	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D		
270.ERCC3	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B		
271.ERCC4	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F		
272.ERCC5	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G		
273.ERCC6	Cockayne syndrome type B		
	COFS syndrome 1		
274.ERCC8	Cockayne syndrome type A		
275.ESCO2	Roberts syndrome		
276.ESPN	Deafness, autosomal recessive 36		
	Deafness, neurosensory, without vestibular involvement, autosomal dominant		
277.ESRRB	Deafness, autosomal recessive 35		
278.ETFA	Glutaric acidemia type 2 (gene ETFA)		
279..ETFB	Glutaric acidemia type 2 (gene ETFB)		
280.ETFDH	Glutaric acidemia type 2 (gene ETFDH)		
281.ETHE1	Ethylmalonic encephalopathy		
282.EVC	Ellis-van Creveld syndrome		
	Weyers acrodental dysostosis		
283.EVC2	Ellis-van Creveld syndrome		
284.EXOSC3	Pontocerebellar hypoplasia, type 1B		
285.EYS	Retinitis pigmentosa 25		
286.F11	Factor XI deficiency, autosomal dominant		
	Factor XI deficiency, autosomal recessive		
287.F2	Dysprothrombinemia		
	Hypoprothrombinemia		
	Pregnancy loss, recurrent, susceptibility to, 2		
	Stroke, ischemic, susceptibility to		
	Thrombophilia due to thrombin defect		
288.F5	Budd-Chiari syndrome		
	Factor V deficiency		
	Pregnancy loss, recurrent, susceptibility to, 1		
	Stroke, ischemic, susceptibility to		
	Thrombophilia due to activated protein C resistance		
	Thrombophilia, susceptibility to, due to factor V Leiden		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
289.F8	Hemophilia A		
290..F9	Hemophilia B		
291.FAH	Tyrosinemia type 1		
292.FAM126A	Hypomyelination - congenital cataract		
293.FAM161A	Retinitis pigmentosa 28		
294.FAM20C	Lethal osteosclerotic bone dysplasia		
295.FANCA	Fanconi anemia, complementation group A		
296.FANCB	Fanconi anemia, complementation group B		
297.FANCC	Fanconi anemia complementation group C		
298.FANCD2	Fanconi anemia, complementation group D2		
299.FANCE	Fanconi anemia, complementation group E		
300.FANCG	Fanconi anemia, complementation group G		
301. FANCI	Fanconi anemia, complementation group I		
302. FANCL	Fanconi anemia, complementation group L		
303.FANCM	Premature ovarian failure 15		
304.FAS	Spermatogenic failure 28		
	Autoimmune lymphoproliferative syndrome, type IA		
305.FASLG	Autoimmune lymphoproliferative syndrome, type IB		
306.FASTKD2	Mitochondrial complex IV deficiency		
307.FBLN5	Cutis laxa, autosomal dominant 2		
	Cutis laxa, autosomal recessive, type IA		
	Macular degeneration, age-related, 3		
308.FERMT3	Leukocyte adhesion deficiency, type III		
309.FGA	Congenital fibrinogen deficiency (gene FGA)		
310.FGB	Afibrinogenemia, congenital/Hypofibrinogenemia, congenital		
	Dysfibrinogenemia, congenital		
311.FGD1	Aarskog-Scott syndrome		
	Mental retardation, X-linked syndromic 16		
312.FGD4	Charcot-Marie-Tooth disease type 4H		
313.FH	Fumaric aciduria		
314.FHL1	Uruguay faciocardiomusculoskeletal syndrome		
	Emery-Dreifuss muscular dystrophy 6, X-linked		
	Myopathy, X-linked, with postural muscle atrophy		
	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset		
	Reducing body myopathy, X-linked 1b, with late childhood or adult onset		
	Scapuloperoneal myopathy, X-linked dominant		
315.FIG4	Amyotrophic lateral sclerosis 11		
	Charcot-Marie-Tooth disease, type 4J		
	Polymicrogyria, bilateral temporooccipital		
	Yunis-Varon syndrome		
316. FKRP	Autosomal recessive limb-girdle muscular dystrophy type 2I		
	Congenital muscular dystrophy type 5B		
	Muscle-eye-brain disease		
317. FKTN	Autosomal recessive limb-girdle muscular dystrophy type 2M		
	Congenital muscular dystrophy type 4B		
	Fukuyama congenital muscular dystrophy		

GENE	MALATTIA	<pid>	<pid>P
318.FLNA	Cardiac valvular dysplasia, X-linked		
	Congenital short bowel syndrome		
	FG syndrome 2		
	Frontometaphyseal dysplasia 1		
	Heterotopia, periventricular, 1		
	Intestinal pseudoobstruction, neuronal		
	Melnick-Needles syndrome		
	Otopalatodigital syndrome, type I		
	Otopalatodigital syndrome, type II		
	Terminal osseous dysplasia		
319.FLVCR1	Ataxia, posterior column, with retinitis pigmentosa		
320.FMR1	Fragile X syndrome		
	Fragile X tremor/ataxia syndrome		
	Premature ovarian failure 1		
321.FOLR1	Neurodegeneration due to cerebral folate transport deficiency		
322.FOXG1	Rett syndrome, congenital variant		
323.FOXN1	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy		
324.FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked		
325.FRAS1	Fraser syndrome (gene FRAS1)		
326.FREM2	Fraser syndrome (gene FRAS2)		
32.7.FTCD	Glutamate formiminotransferase deficiency		
328.FTSJ1	Mental retardation, X-linked 9		
329.FUCA1	Fucosidosis		
330.FXN	Friedreich ataxia		
	Friedreich ataxia with retained reflexes		
331. G6PC	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a		
332 6PC3	Síndrome de Dursun		
323.G6PD	Favism		
	Hemolytic anemia due to G6PD deficiency		
334.GAA	Glycogen storage disease due to acid maltase deficiency		
335.GALC	Krabbe disease		
336.GALE	Galactose epimerase deficiency		
337.GALK1	Galactokinase deficiency with cataracts		
338.GALNS	Mucopolysaccharidosis IVA		
339.GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1		
340.GALT	Classic galactosemia		
341.GAMT	Guanidinoacetate methyltransferase deficiency		
342.GAN	Giant axonal neuropathy-1		
343.GBA	Fetal Gaucher disease		
	Gaucher disease type 2		
	Gaucher disease type 3		
	Gaucher disease type 3C		
344.GBE1	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form		
345.GCDH	Glutaryl-CoA dehydrogenase deficiency		
346.GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia		
	Hyperphenylalaninemia, BH4-deficient, B		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
347.GCSH	Glycine encephalopathy		
348.GDAP1	Autosomal dominant Charcot-Marie-Tooth disease type 2K		
	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness		
	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A		
	Charcot-Marie-Tooth disease type 4A		
349.GDF5	Acromesomelic dysplasia, Hunter-Thompson type		
	Acromesomelic dysplasia, Hunter-Thompson type		
	Brachydactyly, type A2		
	Brachydactyly, type C		
	Chondrodysplasia, Grebe type		
	Du Pan syndrome		
	Multiple synostoses syndrome 2		
	Osteoarthritis-5		
	Symphalangism, proximal, 1B		
350.GDI1	Mental retardation, X-linked 41		
351.GFM1	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1		
352.GHRHR	Growth hormone deficiency, isolated, type IV		
353.GJA1	Atrioventricular septal defect 3		
	Craniometaphyseal dysplasia, autosomal recessive		
	Erythrokeratodermia variabilis et progressiva 3		
	Hypoplastic left heart syndrome 1		
	Oculodentodigital dysplasia		
	Oculodentodigital dysplasia, autosomal recessive		
	Palmoplantar keratoderma with congenital alopecia		
	Syndactyly, type III		
354.GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1		
355.GJB2	Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A (gene GJB2)		
355.GJB3	Deafness, autosomal dominant 2B		
	Deafness, autosomal dominant, with peripheral neuropathy		
	Deafness, autosomal recessive		
	Deafness, digenic, GJB2/GJB3		
	Erythrokeratodermia variabilis et progressiva 1		
357.GJB6	Deafness, autosomal dominant 3B		
	Deafness, autosomal recessive 1B		
	Deafness, digenic GJB2/GJB6		
	Ectodermal dysplasia 2, Clouston type		
358.GJC2	Pelizaeus-Merzbacher-like due to GJC2 mutation		
359.GK	Glycerol kinase deficiency		
360.GLA	Fabry disease		
361.GLB1	GM1 gangliosidosis type 1		
	GM1 gangliosidosis type 2		
	GM1 gangliosidosis type 3		
	Mucopolysaccharidosis type 4B		
362.GLDC	Glycine encephalopathy		
363.GLE1	Lethal congenital contracture syndrome type 1		
364.GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism		
365.GM2A	GM2-gangliosidosis, AB variant		

GENE	MALATTIA	<pid>	<pid>P
366.GNAS	ACTH-independent macronodular adrenal hyperplasia		
	McCune-Albright syndrome, somatic, mosaic		
	Osseous heteroplasia, progressive		
	Pituitary adenoma 3, multiple types, somatic		
	Pseudohypoparathyroidism Ia		
	Pseudohypoparathyroidism Ib		
	Pseudohypoparathyroidism Ic		
	Pseudopseudohypoparathyroidism		
367.GNE	Nonaka myopathy		
	Sialuria		
368.GNMT	Glycine N-methyltransferase deficiency		
369.GNPTAB	Mucolipidosis type 2		
	Mucolipidosis type 3		
370.GNPTG	Mucolipidosis III gamma		
371.GNRHR	Fertile eunuch syndrome		
	Hypogonadotropic hypogonadism 7 without anosmia		
372.GNS	Mucopolysaccharidosis type IIID		
373. GORAB	Bernard-Soulier syndrome, type A1 (recessive)		
	Bernard-Soulier syndrome, type A2 (dominant)		
	Nonarteritic anterior ischemic optic neuropathy, susceptibility to		
	von Willebrand disease, platelet-type		
374.GP1BA	Bernard-Soulier syndrome, type A1 (recessive)		
	Bernard-Soulier syndrome, type A2 (dominant)		
	Nonarteritic anterior ischemic optic neuropathy, susceptibility to		
	von Willebrand disease, platelet-type		
375.GP1BB	Bernard-Soulier syndrome, type B		
	Giant platelet disorder, isolated		
376.GP9	Bernard-Soulier syndrome, type C		
377.GPC3	Simpson-Golabi-Behmel syndrome, type 1		
378.GPR143	Nystagmus 6, congenital, X-linked		
	Ocular albinism, type I, Nettleship-Falls type		
379.GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive		
380.GPR98	Usher syndrome type 2C		
381.GRHPR	Hyperoxaluria, primary, type II		
382.GRIA3	Mental retardation, X-linked 94		
383.GRIK2	Mental retardation, autosomal recessive, 6		
384.GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive		
385.GRXCR1	Deafness, autosomal recessive 25		
386.GSS	Glutathione synthetase deficiency with 5-oxoprolinuria		
387.GTF2H5	Trichothiodystrophy, complementation group A		
388.GUCY2D	Choroidal dystrophy, central areolar 1		
	Cone-rod dystrophy 6		
	Leber congenital amaurosis 1		
389..GUSB	Mucopolysaccharidosis type 7		
390.HADH	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency		
391.HADHA	Mitochondrial trifunctional protein deficiency		
392.HADHB	Mitochondrial trifunctional protein deficiency		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
393.HAL	Histidinemia		
394.HAMP	Histidinemia		
395.HAX1	Neutropenia, severe congenital 3, autosomal recessive		
396.HBA1	Alpha-thalassemia		
397.HBA2	Erythrocytosis 7		
	Heinz body anemia		
	Hemoglobin H disease, deletional and nondeletional		
	Thalassemia, alpha-		
398.HBB	Beta-thalassemia		
	Sickle cell anemia		
399.HCCS	Linear skin defects with multiple congenital anomalies 1		
400.HESX1	Combined pituitary hormone deficiencies, genetic forms		
401.HEXA	Tay-Sachs disease		
402.HEXB	Sandhoff disease		
403. HFE	Alzheimer disease, susceptibility to		
	Hemochromatosis		
	Microvascular complications of diabetes 7		
	Porphyria cutanea tarda, susceptibility to		
	Porphyria variegata, susceptibility to		
	Transferrin serum level QTL2		
	Hemochromatosis, type 2A		
404.HGD	Alkaptonuria		
405.HGF	Deafness, autosomal recessive 39		
406.HGSNAT	Sanfilippo syndrome type C		
407.HIBCH	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency		
408.HLCS	Holocarboxylase synthetase deficiency		
409.HMGCL	3-hydroxy-3-methylglutaric aciduria		
410.HMOX1	Heme oxygenase-1 deficiency		
	Pulmonary disease, chronic obstructive, susceptibility to		
411.HOGA1	Hyperoxaluria, primary, type III		
412.HP	Anhaptoglobinemia		
	Hypohaptoglobinemia		
413.HPD	Tyrosinemia type 3		
414.HPRT1	Kelley-Seegmiller syndrome		
	Lesch-Nyhan syndrome		
415.HPS1	Hermansky-Pudlak syndrome 1		
416.HPS3	Hermansky-Pudlak syndrome 3		
417.HSD11B2	Apparent mineralocorticoid excess		
418.HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency		
419. HSD17B3	Pseudohermaphroditism, male, with gynecomastia		
420.HSD17B4	Bifunctional enzyme deficiency		
	Perrault syndrome		
421.HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency		
422.HSPD1	Leukodystrophy, hypomyelinating, 4		
	Spastic paraplegia 13, autosomal dominant		
423.HSPG2	Schwartz-Jampel syndrome		

GENE	MALATTIA	<pid>	<pid>P
424. TRA1	CARASIL syndrome		
	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2		
	Macular degeneration, age-related, 7		
	Macular degeneration, age-related, neovascular type		
425.HUWE1	Mental retardation, X-linked syndromic, Turner type		
426.HYAL1	Mucopolysaccharidosis type IX		
427.HYLS1	Hydrolethalus syndrome		
428.ICOS	Immunodeficiency, common variable, 1		
429.DH3B	Retinitis pigmentosa 46		
430.IDS	Mucopolysaccharidosis type 2		
431.IDUA	Mucopolysaccharidosis Ih		
	Mucopolysaccharidosis Ih/s		
	Mucopolysaccharidosis Is		
432.IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR		
433.IFNGR2	Immunodeficiency 28, mycobacteriosis		
434.IFT80	Jeune syndrome		
435.IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia		
436. GF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency		
437.IGHMBP2	Spinal muscular atrophy with respiratory distress		
438.IKBKAP	Familial dysautonomia		
439.IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency		
	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency		
	Immunodeficiency 33		
	Incontinentia pigmenti, type II		
440.IL12B	Immunodeficiency 29, mycobacteriosis		
441.IL12RB1	Immunodeficiency 30		
442.IL1RAPL1	Mental retardation, X-linked 21/34		
443.IL1RN	Interleukin 1 receptor antagonist deficiency		
444.IL2RA	Diabetes, mellitus, insulin-dependent, susceptibility to, 10		
	Immunodeficiency 41 with lymphoproliferation and autoimmunity		
445.IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency		
446.IMPDH1	Leber congenital amaurosis 11		
	Retinitis pigmentosa 10		
447.IMPG2	Macular dystrophy, vitelliform, 5		
	Retinitis pigmentosa 56		
448.INSR	Leprechaunism		
449.INVS	Nephronophthisis 2, infantile		
450.IQCB1	Senior-Loken syndrome 5		
451.IQSEC2	Mental retardation, X-linked 1/78		
452.ISCU	Myopathy with lactic acidosis, hereditary		
453.ITGA6	Junctional epidermolysis bullosa - pyloric atresia		
454.ITGB4	Junctional epidermolysis bullosa with piloric atresia		
	Junctional epidermolysis bullosa, non-Herlitz type		
455.IVD	Isovaleric acidemia		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
456. IYD	Thyroid dyshormonogenesis 4		
457.AK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency		
458.KCNJ1	Antenatal Bartter syndrome		
459.KCNJ11	Diabetes mellitus, transient neonatal, 3		
	Diabetes mellitus, type 2, susceptibility to		
	Diabetes, permanent neonatal, with or without neurologic features		
	Hyperinsulinemic hypoglycemia, familial, 2		
	Maturity-onset diabetes of the young, type 13		
460.KCNJ13	Leber congenital amaurosis 16		
	Snowflake vitreoretinal degeneration		
461.KCNV2	Retinal cone dystrophy 3B		
462.KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type		
463..KIAA2022	Mental retardation, X-linked 98		
464.KIF7	Acrocallosal syndrome		
	Al-Gazali-Bakalinova syndrome		
	Hydrolethalus syndrome 2		
	Joubert syndrome 12		
465.L1CAM	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity- hydrocephalus syndrome		
	Masa syndrome		
466.LAMA2	Congenital muscular dystrophy type 1A		
467.LAMA3	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)		
	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)		
	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMA3)		
468.LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities		
	Pierson syndrome		
469.LAMB3	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMB3)		
470.LAMC2	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2		
	unctional epidermolysis bullosa, non-Herlitz type (gene LAMC2)		
471.LAMP2	Danon disease		
472..LARGE	Congenital muscular dystrophy type 1D		
	Muscle-eye-brain disease		
473.LBR	Greenberg dysplasia		
474.LCA5	Leber congenital amaurosis 5		
475.LDHA	Glycogen storage disease XI		
476.LDLR	Hypercholesterolemia, familial, 1		
	LDL cholesterol level QTL2		
477.LDLRAP1	Hypercholesterolemia, familial, 4		
478.LEPRE1	Osteogenesis imperfecta type 8		

GENE	MALATTIA	<pid>	<pid>P
479. LHCGR	Leydig cell adenoma, somatic, with precocious puberty		
	Leydig cell hypoplasia with hypergonadotropic hypogonadism		
	Leydig cell hypoplasia with pseudohermaphroditism		
	Luteinizing hormone resistance, female		
	Precocious puberty, male		
480.LHFPL5	Deafness, autosomal recessive 67		
481.LHX3	Combined pituitary hormone deficiency with spine abnormalities		
482.LIFR	Stüve-Wiedemann syndrome		
483.LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation		
484.LIPA	Cholesteryl ester storage disease		
	Wolman disease		
485.LIPH	Hypotrichosis 7		
	Woolly hair, autosomal recessive 2 with or without hypotrichosis		
486.LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type		
487.LMNA	Charcot-Marie-Tooth disease axonal type 2B1		
	Lethal restrictive dermopathy		
	Mandibuloacral dysplasia with type A lipodystrophy		
488.LOXHD1	Deafness, autosomal recessive 77		
489.LPL	Combined hyperlipidemia, familial		
	High density lipoprotein cholesterol level QTL 11		
	Lipoprotein lipase deficiency		
490.LRAT	Leber congenital amaurosis 14		
491.LRP2	Retinal dystrophy, early-onset severe		
	Retinitis pigmentosa, juvenile		
	Donnai-Barrow syndrome		
492.LRP5	Bone mineral density variability 1		
	Exudative vitreoretinopathy 4		
	Hyperostosis, endosteal		
	Osteopetrosis, autosomal dominant 1		
	Osteoporosis		
	Osteoporosis-pseudoglioma syndrome		
	Osteosclerosis		
	Polycystic liver disease 4 with or without kidney cysts		
	van Buchem disease, type 2		
493.LRPPRC	French-Canadian type Leigh syndrome		
494.LRTOMT	Deafness, autosomal recessive 63		
495.LYST	Chediak-Higashi syndrome		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
496. AGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia		
497.MAK	Retinitis pigmentosa 62		
498.MAN2B1	Mannosidosis, alpha-, types I and II		
499.MARVEL D2	Deafness, autosomal recessive 49		
500.MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency		
	Methionine adenosyltransferase deficiency, autosomal recessive		
501.MATN3	Epiphyseal dysplasia, multiple, 5		
	Osteoarthritis susceptibility 2		
	Spondyloepimetaphyseal dysplasia		
502.MBTPS2	Ichthyosis follicularis - alopecia - photophobia		
503.MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency		
504.MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency		
505.MCEE	Methylmalonyl-CoA epimerase deficiency		
506.MCOLN1	Mucolipidosis type 4		
507.MCPH1	Microcephaly 1, primary, autosomal recessive		
508.MECP2	Severe neonatal-onset encephalopathy with microcephaly		
509.MED12	X-linked intellectual deficit with marfanoid habitus		
510.MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy		
511.MED25	Basel-Vanagait-Smirin-Yosef syndrome		
	Charcot-Marie-Tooth disease, type 2B2		
512.MEFV	Familial Mediterranean fever		
513.MERTK	Retinitis pigmentosa 38		
514.MESP2	Spondylocostal dysostosis 2, autosomal recessive		
515.MFRP	Microphthalmia, isolated 5		
516.MFSD8	Nanophthalmos 2		
	Nanophthalmos 2		
	Late infantile neuronal ceroid lipofuscinosis		
517.MGAT2	Congenital disorder of glycosylation type 2a		
518.MID1	Opitz GBBB syndrome, type I		
519.MKKS	Bardet-Biedl syndrome 6		
520.MKS1	McKusick-Kaufman syndrome		
	McKusick-Kaufman syndrome		
	Meckel syndrome type 1		
521.MLC1	Megalencephalic leukoencephalopathy with subcortical cysts		
522 MLYCD	Malonyl-CoA decarboxylase deficiency		
523.MMAA	Vitamin B12-responsive methylmalonic acidemia type cblA		
524.MMAB	Vitamin B12-responsive methylmalonic acidemia type cblB		
525.MMACHC	Methylmalonic acidemia with homocystinuria, type cblC		
	Methylmalonic acidemia with homocystinuria, type cblD		
526.MMADHC	Homocystinuria, cblD type, variant 1		
527.MOCS1	Methylmalonic aciduria and homocystinuria, cblD type		
	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS1)		

GENE	MALATTIA	<pid>	<pid>P
528.MOCS2	Methylmalonic aciduria and homocystinuria, cblD type		
	Methylmalonic aciduria, cblD type, variant 2		
	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS2)		
529.MOGS	Congenital disorder of glycosylation, type IIb		
530.MPDU1	Congenital disorder of glycosylation, type If		
531.MPI	Congenital disorder of glycosylation type 1b		
532.MPL	Thrombocythemia 2		
	Thrombocytopenia, congenital amegakaryocytic		
533.MPV17	Methylmalonic aciduria, cblD type, variant 2		
	Navajo neurohepatopathy		
534.MPZ	Charcot-Marie-Tooth disease, type 1B		
	Charcot-Marie-Tooth disease, type 2I		
	Charcot-Marie-Tooth disease, type 2J		
	Dejerine-Sottas disease		
	Neuropathy, congenital hypomyelinating		
	Roussy-Levy syndrome		
535.MRE11	Ataxia-telangiectasia-like disorder 1		
536.MRPS16	Combined oxidative phosphorylation defect type 2		
537.MRPS22	Combined oxidative phosphorylation defect type 5		
538.MTHFR	Homocystinuria due to MTHFR deficiency		
539.MTM1	X-linked centronuclear myopathy		
540. MTMR2	Charcot-Marie-Tooth disease, type 4B1		
541.MTR	Homocystinuria-megaloblastic anemia, cblG complementation type		
	Neural tube defects, folate-sensitive, susceptibility to		
542.MTRR	Homocystinuria-megaloblastic anemia, cbl E type		
	Neural tube defects, folate-sensitive, susceptibility to		
543.MTTP	Abetalipoproteinemia		
544.MUT	Metabolic syndrome, protection against		
	Vitamin B12-unresponsive methylmalonic acidemia type mut-		
545.MVK	Mevalonic aciduria		
546.MYD88	Macroglobulinemia, Waldenstrom		
	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency		
547.MYO15A	Deafness, autosomal recessive 3		
548.MYO3A	Deafness, autosomal recessive 30		
549.MYO5A	Griscelli disease type 1		
550.MYO6	Deafness, autosomal dominant 22		
551.MYO7A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB2		
	Usher syndrome type 1		
552.NAGA	Kanzaki disease		
553.NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
554.NAGS	Hyperammonemia due to N-acetylglutamate synthetase deficiency		
555.NBN	Aplastic anemia		
	Nijmegen breakage syndrome		
556.NDP	Exudative vitreoretinopathy 2, X-linked		
	Norrie disease		
557.NDRG1	Charcot-Marie-Tooth disease, type 4D		
558.NDUFA1	Mitochondrial complex I deficiency, nuclear type 12		
	Mitochondrial complex I deficiency		
559. NDUFA7	Mitochondrial complex I deficiency, nuclear type 12		
560. NDUFAF2	Mitochondrial complex I deficiency, nuclear type 1		
	Leigh syndrome		
	Mitochondrial complex I deficiency		
561.NDUFAF4	Mitochondrial complex I deficiency		
562.NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16		
563.NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency		
	Mitochondrial complex I deficiency		
564.NDUFS4	Leigh syndrome		
	Mitochondrial complex I deficiency		
565.NDUFS5	NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 5		
566.NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of		
567.NDUFS7	Leigh syndrome		
568.NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency		
569.NDUFV1	Mitochondrial complex I deficiency		
570.NEB	Nemaline myopathy 2		
571.NEFL	Charcot-Marie-Tooth disease, dominant intermediate G		
572.NEU. 1	Sialidosis, type I		
	Sialidosis, type II		
573. NEUROG 3	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells		
574.NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation		
575.NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora)		
576.NHP2	Dyskeratosis congenita, autosomal recessive 2		
577.NHS	Cataract 40, X-linked		
	Nance-Horan syndrome		
578.NKX2-1	Chorea, hereditary benign		
	Choreoathetosis, hypothyroidism, and neonatal respiratory distress		
	Thyroid cancer, nonmedullary, 1		
579.NKX2-5	Atrial septal defect 7, with or without AV conduction defects		
	Conotruncal heart malformations, variable		
	Hypoplastic left heart syndrome 2		
	Hypothyroidism, congenital nongoitrous, 5		
	Tetralogy of Fallot		
580.NKX2-5	Ventricular septal defect 3		

GENE	MALATTIA	<pid>	<pid>P
581.NLGN3	Asperger syndrome susceptibility, X-linked 1		
	Autism susceptibility, X-linked 1		
582.NLGN4X	Mental retardation, X-linked		
583.NLRP7	Hydatidiform mole, recurrent, 1		
584.NMNAT1	Leber congenital amaurosis 9		
585.NOP10	Dyskeratosis congenita, autosomal recessive 1		
586.NPC1	Niemann-Pick disease type C1		
587.NPC2	Niemann-Pick disease type C2		
588.NPHP1	Joubert syndrome 4		
589.NPHP3	Renal-hepatic-pancreatic dysplasia		
	Senior-Loken syndrome 1		
590.NPHP4	Senior-Loken syndrome		
591.NPHS1	Nephrotic syndrome, type 1		
592.NPHS2	Nephrotic syndrome, type 2		
593.NR0B1	46XY sex reversal 2, dosage-sensitive		
594.NR2E3	Enhanced S-cone syndrome		
	Retinitis pigmentosa 37		
595.NR5A1	46XY sex reversal 3		
	Adrenocortical insufficiency		
596.NSD1	Beckwith-Wiedemann syndrome		
	Sotos syndrome 1		
597.NSDHL	CHILD syndrome		
	CK syndrome		
598.NSUN2	Mental retardation, autosomal recessive 5		
599.NTRK1	Hereditary sensory and autonomic neuropathy type 4		
600.NUP62	Infantile bilateral striatal necrosis		
601.NXF5	NUCLEAR RNA EXPORT FACTOR 5		
602.NYX	Night blindness, congenital stationary (complete), 1A, X-linked		
603.OAT	Gyrate atrophy of choroid and retina with or without ornithinemia		
604.OCA2	Albinism, brown oculocutaneous		
	Albinism, oculocutaneous, type II		
	Skin/hair/eye pigmentation 1, blond/brown hair		
	Skin/hair/eye pigmentation 1, blue/nonblue eyes		
605.OCRL	Dent disease 2Oc		
	Oculocerebrorenal syndrome		
606.OFD1	Simpson-Golabi-Behmel syndrome type 2		
607.OPA3	3-methylglutaconic aciduria type 3		
608.OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance		
609.ORAI1	Immunodeficiency 9		
	Myopathy, tubular aggregate, 2		
610.OSTM1	Osteopetrosis, autosomal recessive 5		
611.OTC	Ornithine transcarbamylase deficiency		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
612.OTOA	Deafness, autosomal recessive 22		
613.OTO F	Auditory neuropathy, autosomal recessive, 1		
614.OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency		
615.PAH	Phenylketonuria		
616.PAK3	Mental retardation, X-linked 30/47		
617.PALB2	Fanconi anemia, complementation group N		
618.PANK2	Pantothenate kinase-associated neurodegeneration		
619.PAX3	Craniofacial-deafness-hand syndrome		
	Rhabdomyosarcoma 2, alveolar		
	Waardenburg syndrome, type 1		
	Waardenburg syndrome, type 3		
620.PAX6	Aniridia		
	Anterior segment dysgenesis 5, multiple subtypes		
	Cataract with late-onset corneal dystrophy		
	Coloboma of optic nerve		
	Coloboma, ocular		
	Foveal hypoplasia 1		
	Keratitis		
	Morning glory disc anomaly		
	Optic nerve hypoplasia		
621.PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia		
622.PC	Pyruvate carboxylase deficiency		
623.PCBD1	Hyperphenylalaninemia, BH4-deficient, D		
624.PCCA	Propionic acidemia (gene PCCA)		
625.PCCB	Propionic acidemia (gene PCCB)		
626.PCDH15	Deafness, autosomal recessive 23		
	Usher syndrome, type 1D/F digenic		
	Usher syndrome, type 1F		
627.PCDH19	Epileptic encephalopathy, early infantile, 9		
628.PDE6A	Retinitis pigmentosa 43		
629.PDE6B	Night blindness, congenital stationary, autosomal dominant 2		
	Retinitis pigmentosa-40		
630.PDE6C	Cone dystrophy 4		
631.PDE6G	Retinitis pigmentosa 57		
632.PDHA1	Leigh syndrome, X-linked		
633.PDHB	Pyruvate dehydrogenase E1-beta deficiency		
634.PDHX	Lacticacidemia due to PDX1 deficiency		
635.PDP1	Pyruvate dehydrogenase phosphatase deficiency		
636.PDSS1	Deafness - encephaloneuropathy - obesity - valvulopathy		
637.PDSS2	Leigh syndrome with nephrotic syndrome		
638.PDX1	Diabetes mellitus, type II, susceptibility to		
	MODY, type IV		
	Pancreatic agenesis 1		
639.PDZD7	Deafness, autosomal recessive 57		
	Retinal disease in Usher syndrome type IIA, modifier of		
	Usher syndrome, type IIC, GPR98/PDZD7 digenic		

GENE	MALATTIA	<pid>	<pid>P
640.PEPD	Prolidase deficiency		
641.PEX1	Zellweger syndrome 1A		
642.PEX10	Peroxisome biogenesis disorder 6A (Zellweger)		
	Peroxisome biogenesis disorder 6B		
643.PEX12	Neonatal adrenoleukodystrophy (gene PEX12)		
644.PEX13	Peroxisome biogenesis disorder 11A (Zellweger)		
	Peroxisome biogenesis disorder 11B		
645.PEX2	Peroxisome biogenesis disorder 5A (Zellweger)		
646.PEX26	Peroxisome biogenesis disorder 5B		
	Peroxisome biogenesis disorder 5B		
	Neonatal adrenoleukodystrophy (gene PEX26)		
	Zellweger syndrome 7A		
647.PEX5	Neonatal adrenoleukodystrophy (gene PEX5)		
648.PEX6	Heimler syndrome 2		
649.PEX7	Peroxisome biogenesis disorder 4A (Zellweger)		
	Peroxisome biogenesis disorder 4B		
	Rhizomelic chondrodysplasia punctata type 1		
650.PFKM	Glycogen storage disease VII		
651.PGK1	Phosphoglycerate kinase 1 deficiency		
652 PGM1	Congenital disorder of glycosylation, type It		
653.PHF8	Mental retardation syndrome, X-linked, Siderius type		
654.PHGDH	Neu-Laxova syndrome 1		
	Phosphoglycerate dehydrogenase deficiency		
655.PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency		
	Glycogen storage disease IXc		
656.PHYH	Refsum disease		
657.PKHD1	Autosomal recessive polycystic kidney disease		
658.PKLR	Hemolytic anemia due to red cell pyruvate kinase deficiency		
659.PLA2G6	Infantile neuroaxonal dystrophy 2A		
	Infantile neuroaxonal dystrophy 2B		
660.PLCE1	Nephrotic syndrome, tupe 3		
661.PLEC	Epidermolysis bullosa simplex with muscular dystrophy		
	Epidermolysis bullosa simplex with pyloric atresia		
	Limb girdle dystrophy with epidermolysis bullosa simplex		
662.PLEKHG5	Autosomal recessive distal spinal muscular atrophy type 4		
663.PLG	Plasminogen deficiency type 1		
664.PLOD1	Ehlers-Danlos syndrome type 6		
665.PLP1	Spastic paraplegia type 2, X-linked		
666.PMM2	Congenital disorder of glycosylation type 1a		
667.PMP22	Charcot-Marie-Tooth disease, type 1A		
	Charcot-Marie-Tooth disease, type 1E		
	Dejerine-Sottas disease		
	Roussy-Levy syndrome		
668.PNPO	Pyridoxal phosphate-responsive seizures		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
669.POLG	Alpers syndrome		
	Autosomal recessive progressive external ophthalmoplegia		
	Mitochondrial neurogastrointestinal encephalomyopathy		
	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis		
670.POLR1C	Leukodystrophy, hypomyelinating, 11		
671.POMGNT 1	Treacher Collins syndrome 3		
	Treacher Collins syndrome 3		
	Autosomal recessive limb-girdle muscular dystrophy type C		
	Congenital muscular dystrophy with cerebellar involvement		
	Walker-Warburg syndrome (gene POMGNT1)		
672.POMT1	Autosomal recessive limb-girdle muscular dystrophy type C		
	Congenital muscular dystrophy with cerebellar involvement		
	Walker-Warburg syndrome (gene POMT1)		
673.POMT2	Autosomal recessive limb-girdle muscular dystrophy type C		
	Congenital muscular dystrophy with cerebellar involvement		
	Walker-Warburg syndrome (gene POMT2)		
674.POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis		
675.POU1F1	Combined pituitary hormone deficiencies, genetic forms		
676.POU3F4	Deafness, X-linked 2		
677.PPT1	Adult neuronal ceroid lipofuscinosis		
678.PQBP1	Renpenning syndrome		
679.PRCD	Retinitis pigmentosa 36		
680.PRF1	Hemophagocytic lymphohistiocytosis, familial, 2		
681.PRKRA	Dystonia 16		
682.PRODH	Hyperprolinemia, type I		
	Schizophrenia, susceptibility to, 4		
683.PROM1	Cone-rod dystrophy 12		
684.PROP1	Macular dystrophy, retinal, 2		
	Combined pituitary hormone deficiencies, genetic forms		
685.PRPS1	Retinitis pigmentosa 41		
	Lethal ataxia with deafness and optic atrophy		
	X-linked Charcot-Marie-Tooth disease type 5		
686.PRSS12	Macular dystrophy, retinal, 2		
	Retinitis pigmentosa 41		
	Stargardt disease 4		
	Stargardt disease 4		
	Mental retardation, autosomal recessive 1		
687.PRX	Charcot-Marie-Tooth disease type 4F		
688.PSAP	Encephalopathy due to prosaposin deficiency		
	Krabbe disease		
	Metachromatic leukodystrophy		

GENE	MALATTIA	<pid>	<pid>P
689.PSAT1	Phosphoserine aminotransferase deficiency		
690.PTEN	Neu-Laxova syndrome 2		
	Bannayan-Riley-Ruvalcaba syndrome		
	Cowden syndrome 1		
	Lhermitte-Duclos syndrome		
	Macrocephaly/autism syndrome		
691.PTH1R	Chondrodysplasia, Blomstrand type		
	Eiken syndrome		
	Failure of tooth eruption, primary		
	Metaphyseal chondrodysplasia, Murk Jansen type		
692.PTS	Hyperphenylalaninemia, BH4-deficient, A		
693.PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1		
694.PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency		
695.QDPR	Hyperphenylalaninemia, BH4-deficient, C		
696.RAB23	Carpenter syndrome		
697.RAB27A	Griscelli disease type 2		
698.RAB39B	Mental retardation, X-linked 72		
699.RAB3GAP 1	Micro syndrome		
700.RAB3GAP 2	Cataract - intellectual deficit - hypogonadism		
701.RAD51C	Fanconi anemia, complementation group O		
702.RAG1	Breast-ovarian cancer, familial, susceptibility to, 3		
	Breast-ovarian cancer, familial, susceptibility to, 3		
	Combined immunodeficiency with skin granulomas		
	Omenn syndrome (gene RAG1)		
	Severe combined immunodeficiency due to complete RAG1/2 deficiency		
703.RAG2	Combined immunodeficiency with skin granulomas		
	Omenn syndrome (gene RAG2)		
	Severe combined immunodeficiency due to complete RAG1/2 deficiency		
704.RAPSN	Fetal akinesia deformation sequence		
705.RARS2	Pontocerebellar hypoplasia, type 6		
706.RAX	Microphthalmia, isolated 3		
707.RDH12	Leber congenital amaurosis 13		
708.RDX	Deafness, autosomal recessive 24		
709.RELN	Lissencephaly syndrome, Norman-Roberts type		
710.REN	Hyperproreninemia		
	Hyperuricemic nephropathy, familial juvenile 2		
	Renal tubular dysgenesis		
711.RFT1	Congenital disorder of glycosylation, type In		
712.RGR	Retinitis pigmentosa		
713.RHO	Night blindness, congenital stationary, autosomal dominant 1		
	Retinitis pigmentosa 4, autosomal dominant or recessive		
	Retinitis punctata albescens		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
714.RLBP1	Bothnia retinal dystrophy		
	Fundus albipunctatus		
	Newfoundland rod-cone dystrophy		
	Retinitis punctata albescens		
715.RMRP	Anauxetic dysplasia		
	Cartilage-hair hypoplasia		
	Metaphyseal dysplasia without hypotrichosis		
716.RNASEH2 A	Aicardi-Goutieres syndrome 4		
717.RNASEH2 B	Aicardi-Goutieres syndrome 2		
718.RNASEH2 C	Aicardi-Goutieres syndrome 3		
719.RP2	Retinitis pigmentosa 2		
720.RPE65	Leber congenital amaurosis 2		
	Retinitis pigmentosa 20		
721.RPGR	Cone-rod dystrophy, X-linked, 1		
	Macular degeneration, X-linked atrophic		
	Retinitis pigmentosa 3		
	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness		
722.RPGRIP1L	Joubert syndrome with hepatic defect		
	Meckel syndrome, type 5		
723.RPL10	Autism, susceptibility to, X-linked 5		
724.RPS6KA3	Coffin-Lowry syndrome		
	Mental retardation, X-linked 19		
725.RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)		
	Mitochondrial DNA depletion syndrome 8B (MNGIE type)		
726.RS1	Retinoschisis		
727.RYR1	Central core disease		
	King-Denborough syndrome		
	Malignant hyperthermia susceptibility 1		
	Minicore myopathy with external ophthalmoplegia		
	Neuromuscular disease, congenital, with uniform type 1 fiber		
728.SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay		
729.SAG	Oguchi disease-1		
	Retinitis pigmentosa 47		
730.SAMD9	MIRAGE syndrome		
	Tumoral calcinosis, familial, normophosphatemic		
731.SAMHD1	Aicardi-Goutieres syndrome 5		
	Chilblain lupus 2		
732.SBDS	Shwachman-Diamond syndrome		
733.SBF2	Charcot-Marie-Tooth disease, type 4B2		

GENE	MALATTIA	<pid>	<pid>P
734.SC5DL	Lathosterolosis		
735.SCN2A	Epileptic encephalopathy, early infantile, 11		
	Seizures, benign familial infantile, 3		
736.SCNN1A	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1A)		
737.SCNN1B	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1B)		
738.SCNN1G	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1G)		
739.SCO1	Mitochondrial complex IV deficiency		
740.SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1		
741.SEMA4A	Cone-rod dystrophy 10		
	Retinitis pigmentosa 35		
742.SEPN1	Rigid spine syndrome		
743.SEPSECS	Pontocerebellar hypoplasia type 2D		
744.SERPINA1	Emphysema due to AAT deficiency		
	Emphysema-cirrhosis, due to AAT deficiency		
	Hemorrhagic diathesis due to antithrombin Pittsburgh		
	Pulmonary disease, chronic obstructive, susceptibility to		
745.SETX	Amyotrophic lateral sclerosis 4, juvenile		
	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2		
746.SFTPB	Surfactant metabolism dysfunction, pulmonary, 1		
747.SFTPC	Surfactant metabolism dysfunction, pulmonary, 2		
748.SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3		
749.SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4		
750.SGCD	Cardiomyopathy, dilated, 1L		
	Muscular dystrophy, limb-girdle, autosomal recessive 6		
751.SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5		
752.SGSH	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)		
753.SH2D1A	X-linked lymphoproliferative disease		
754.SH3TC2	Charcot-Marie-Tooth disease, type 4C		
	Mononeuropathy of the median nerve, mild		
755.SHROOM 4	Stocco dos Santos X-linked mental retardation syndrome		
756.SIL1	Marinesco-Sjögren syndrome		
757.SIX6	Optic disc anomalies with retinal and/or macular dystrophy		
758.SLC12A1	Antenatal Bartter syndrome type 1		
759.SLC12A3	Gitelman syndrome		
760.SLC12A6	Corpus callosum agenesis - neuronopathy		
761.SLC16A2	Allan-Herndon-Dudley syndrome		
762.SLC17A5	Free sialic acid storage disease, infantile form		
763.SLC19A2	Thiamine-responsive megaloblastic anemia syndrome		
764.SLC22A5	Carnitine deficiency, systemic primary		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
765.SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive		
766.SLC25A13	Citrullinemia, adult-onset type II		
	Citrullinemia, type II, neonatal-onset		
767.SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria		
768.SLC25A20	Carnitine-acylcarnitine translocase deficiency		
769.SLC25A22	Early infantile epileptic encephalopathy		
770.SLC26A2	Achondrogenesis type 1B		
	Atelosteogenesis type II		
	Diastrophic dwarfism		
	Multiple epiphyseal dysplasia type 4		
771.SLC26A3	Diarrhea 1, secretory chloride, congenital		
772.SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct		
	Pendred syndrome		
773.SLC26A5	Deafness, autosomal recessive 61		
774.SLC35A1	Congenital disorder of glycosylation type 2f		
775.SLC35C1	Congenital disorder of glycosylation type 2c		
776.SLC35D1	Schneckenbecken dysplasia		
777.SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type b		
778.SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type c		
779.SLC39A4	Acrodermatitis enteropathica		
780.SLC3A1	Cystinuria		
781.SLC45A2	Albinism, oculocutaneous, type IV		
	Skin/hair/eye pigmentation 5, black/nonblack hair		
	Skin/hair/eye pigmentation 5, dark/fair skin		
	Skin/hair/eye pigmentation 5, dark/light eyes		
782.SLC46A1	Folate malabsorption, hereditary		
783.SLC4A11	Congenital hereditary endothelial dystrophy type II		
	Corneal dystrophy - perceptive deafness		
784.SLC5A5	Folate malabsorption, hereditary		
785.SLC6A19	Hartnup disorder		
	Hyperglycinuria		
	Iminoglycinuria, digenic		
786.SLC6A8	X-linked creatine transporter deficiency		
787.SLC7A7	Lysinuric protein intolerance		
788.SLC7A9	Cystinuria		
789.SLC9A6	Mental retardation, X-linked syndromic, Christianson type		
790.SLX4	Fanconi anemia, complementation group P		
791.SMARCAL1	Fanconi anemia, complementation group P		
792.SMN1	Proximal spinal muscular atrophy type 1		
	Proximal spinal muscular atrophy type 2		
	Proximal spinal muscular atrophy type 3		
	Proximal spinal muscular atrophy type 4		

GENE	MALATTIA	<pid>	<pid>P
793.SMN2	Spinal muscular atrophy, type III, modifier of		
794.SMPD1	Niemann-Pick disease type A		
	Niemann-Pick disease type B		
795.SMS	Mental retardation, X-linked, Snyder-Robinson type		
796.SNAI2	Piebaldism		
	Waardenburg syndrome, type 2D		
797.SNAP29	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome		
798.SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency		
	Panhypopituitarism, X-linked		
799.SP110	Hepatic venoocclusive disease with immunodeficiency		
800.SPG11	Amyotrophic lateral sclerosis 5, juvenile		
	Charcot-Marie-Tooth disease, axonal, type 2X		
	Spastic paraplegia 11, autosomal recessive		
801.SPG20	Troyer syndrome		
802.SPG7	Spastic paraplegia 7, autosomal recessive		
803.SRD5A2	Pseudovaginal perineoscrotal hypospadias		
804.SRD5A3	Congenital disorder of glycosylation, type Iq		
	Kahrizi syndrome		
805.SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia		
806.ST3GAL3	Epileptic encephalopathy, early infantile, 15		
	Mental retardation, autosomal recessive 12		
807.ST3GAL5	Amish infantile epilepsy syndrome		
808.STAR	Congenital lipoid adrenal hyperplasia		
809.STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant		
	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive		
	Immunodeficiency 31C, autosomal dominant		
810.STIL	Microcephaly 7, primary, autosomal recessive		
811.STIM1	Immunodeficiency 10		
	Myopathy, tubular aggregate, 1		
	Stormorken syndrome		
812.STRA6	Syndromic microphthalmia type 9		
813.STRC	Deafness, autosomal recessive 16		
814.STX11	Hemophagocytic lymphohistiocytosis, familial, 4		
815.STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5		
816.SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)		
817.SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria		
818.SUMF1	Multiple sulfatase deficiency		
819.SUOX	Sulfocysteinuria		
820.SURF1	Leigh syndrome, due to COX deficiency		
821.SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders		
822.SYP	Mental retardation, X-linked 96		

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
823.TAF1	Dystonia-Parkinsonism, X-linked		
	Mental retardation, X-linked, syndromic 33		
824.TAT	Tyrosinemia type 2		
825.TAZ	Barth syndrome		
826.TBCE	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome		
827.TCAP	Cardiomyopathy, hypertrophic, 25		
	Muscular dystrophy, limb-girdle, autosomal recessive 7		
828.TCF4	Pitt-Hopkins syndrome		
829.TCIRG1	Autosomal recessive malignant osteopetrosis 1		
830.TCN2	Transcobalamin II deficiency		
831.TECTA	Deafness, autosomal dominant 8/12		
	Deafness, autosomal recessive 21		
832TERT	Dyskeratosis congenita, autosomal dominant 2		
	Dyskeratosis congenita, autosomal recessive 4		
	Leukemia, acute myeloid		
	Melanoma, cutaneous malignant, 9		
	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1		
833TFR2	Hemochromatosis, type 3		
834.TG	Autoimmune thyroid disease, susceptibility to, 3		
	Thyroid dyshormonogenesis 3		
835.TGM1	Ichthyosis, congenital, autosomal recessive 1		
836.TH	Autosomal recessive dopa-responsive dystonia		
837.THRA	Hypothyroidism, congenital, nongoitrous, 6		
838.THRB	Thyroid hormone resistance		
	Thyroid hormone resistance, autosomal recessive		
	Thyroid hormone resistance, selective pituitary		
839.TIMM8A	Mohr-Tranebjaerg syndrome		
840.TK2	Mitochondrial DNA depletion syndrome, myopathic form		
841.TLR3	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2		
	HIV1 infection, resistance to		
842.TMC1	Deafness, autosomal dominant 36		
	Deafness, autosomal recessive 7		
843.TMEM216	Joubert syndrome 2		
	Meckel syndrome 2		
844.TMEM67	COACH syndrome		
	Joubert syndrome 6		
845.TMIE	Deafness, autosomal recessive 6		
846.TMPRSS3	Deafness, autosomal recessive 8/10		
847.TNFRSF1 1B	Paget disease, juvenile		
848.TNNT1	Nemaline myopathy 5, Amish type		
849.TPO	Thyroid dyshormonogenesis 2A		
850.TPP1	Neuronal ceroid lipofuscinosis 2		

GENE	MALATTIA	<pid>	<pid>P
851.TPRN	Deafness, autosomal recessive 79		
852.TRAPPC9	Mental retardation, autosomal recessive 13		
853.TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness		
854.TREX1	Aicardi-Goutières syndrome		
855.TRIM32	Bardet-Biedl syndrome 11		
	Muscular dystrophy, limb-girdle, autosomal recessive 8		
856.TRIM37	MULIBREY nanism		
857.TRIOBP	Deafness, autosomal recessive 28		
858.TRMU	Deafness, mitochondrial, modifier of		
	Liver failure, transient infantile		
859.TSEN54	Pontocerebellar hypoplasia type 2A		
	Pontocerebellar hypoplasia type 4		
860.TSFM	Fatal mitochondrial disease due to combined oxidative phosphorylationdeficiency 3		
861.TSHB	Isolated thyroid-stimulating hormone deficiency		
862 TSHR	Hyperthyroidism, familial gestational		
	Hyperthyroidism, nonautoimmune		
	Hypothyroidism, congenital, nongoitrous, 1		
	Thyroid adenoma, hyperfunctioning, somatic		
	Thyroid carcinoma with thyrotoxicosis		
863.TSPAN7	Mental retardation, X-linked 58		
864.TSPYL1	Sudden infant death with dysgenesis of the testes syndrome		
865.TTC37	Trichohepatoenteric syndrome 1		
866.TTN	Cardiomyopathy, dilated, 1G		
	Cardiomyopathy, familial hypertrophic, 9		
	Muscular dystrophy, limb-girdle, autosomal recessive 10		
	Myopathy, myofibrillar, 9, with early respiratory failure		
	Salih myopathy		
	Tibial muscular dystrophy, tardive		
867.TTPA	Ataxia with vitamin E deficiency		
868.TUBA1A	Lissencephaly 3		
869. TUFM	Combined oxidative phosphorylation deficiency 4		
870.TULP1	Leber congenital amaurosis 15		
	Retinitis pigmentosa 14		
871.TUSC3	ntal retardation, autosomal recessive 7		
872.TYK2	Immunodeficiency 35		
873.TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)		
874.TYR	Albinism, oculocutaneous, type IA		
	Albinism, oculocutaneous, type IB		
	Melanoma, cutaneous malignant, susceptibility to, 8		
	Skin/hair/eye pigmentation 3, blue/green eyes		
	Skin/hair/eye pigmentation 3, light/dark/freckling skin		
	Waardenburg syndrome/albinism, digenic		

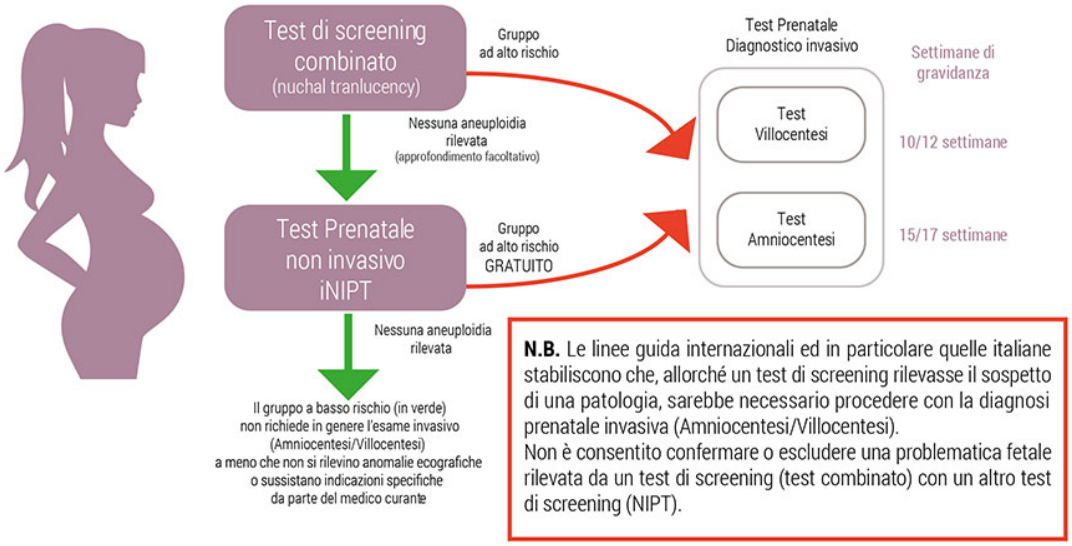
COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	MALATTIA	<pid>	<pid>P
875.TYRP1	Albinism, oculocutaneous, type III		
	Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)		
876.UBA1	X-linked spinal muscular atrophy type 2		
877.UBE2A	Mental retardation, X-linked syndromic, Nascimento-type		
878.UBE3A	Angelman syndrome		
879.UBR1	Johanson-Blizzard syndrome		
880.UGT1A1	Bilirubin, serum level of, QTL1		
	Crigler-Najjar syndrome, type I		
	Crigler-Najjar syndrome, type II		
	Gilbert syndrome		
	Hyperbilirubinemia, familial transient neonatal		
881.UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3		
882.UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1		
883.UPF3B	Mental retardation, X-linked, syndromic 14		
884.UQCRB	Mitochondrial respiratory chain complex III deficiency		
885.UQCRQ	Mitochondrial respiratory chain complex III deficiency		
886.UROS	Porphyria, congenital erythropoietic		
887.USH1C	Autosomal recessive nonsyndromic sensorineural deafness type DFNB18		
	Usher syndrome type 1C		
888.USH1G	Usher syndrome type 1G		
889.USH2A	Usher syndrome type 2A		
890.USP9X	Mental retardation, X-linked 99		
	Mental retardation, X-linked 99, syndromic, female-restricted		
891. VDR	Vitamin D-dependent rickets type 2A		
892.VLDLR	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome		
893.VPS13A	Choreoacanthocytosis		
894.VPS13B	Cohen Syndrome type 1		
895.VPS33B	Arthrogryposis - renal dysfunction - cholestasis		
896.VRK1	Pontocerebellar hypoplasia type 1A		
897.VSX2	Microphthalmia with coloboma 3		
	Microphthalmia, isolated 2		
898.VWF	von Willebrand disease, type 1		
	von Willebrand disease, types 2A, 2B, 2M, and 2N		
	von Willibrand disease, type 3		
899.WAS	Neutropenia, severe congenital, X-linked		
	Thrombocytopenia, X-linked		
	Thrombocytopenia, X-linked, intermittent		
	Wiskott-Aldrich syndrome		
900.WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations		
901.WFS1	Wolfram syndrome 1		
902.WISP3	Arthropathy, progressive pseudorheumatoid, of childhood		
	Spondyloepiphyseal dysplasia tarda with progressive arthropathy		

GENE	MALATTIA	<pid>	<pid>P
903.WNT10A	Odontoonychodermal dysplasia		
	Schopf-Schulz-Passarge syndrome		
	Tooth agenesis, selective, 4		
904.WNT3	Tetra-amelia, autosomal recessive		
905.WNT7A	Aplasia/hypoplasia of limbs and pelvis		
	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly		
906.WRN	Werner syndrome		
907.XIAP	Lymphoproliferative syndrome, X-linked, 2		
908.XPA	Xeroderma pigmentosum complementation group A		
909.XPC	Xeroderma pigmentosum, group C		
910.ZDHHC9	Mental retardation, X-linked syndromic, Raymond type		
911ZEB2	Mowat-Wilson syndrome		
912.ZFYVE6	Spastic paraplegia 15, autosomal recessive		
913.ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked		
	Heterotaxy, visceral, 1, X-linked		
914.ZMPSTE24	Lethal restrictive dermopathy		
	Mandibuloacral dysplasia with type B lipodystrophy		
915. ZNF469	Brittle cornea syndrome		
916. ZNF711	Mental retardation, X-linked 97		

PROTOCOLLO DI INDAGINI PRENATALI NON INVASIVE



INFORMAZIONI SUL TEST: TECNICA, POTENZIALITA' E LIMITI:

Il **FetalDNA - Complete Genetic Scan (CGS)**: è un esame genetico determinato dall'abbinamentodi due protocolli specifici, sul DNA Fetale e sul DNA genomico della coppia di genitori. Per entrambi i protocolli viene impiegata la NGS (Next Generation Sequencing).
Viene impiegata la metodica di NGS (Next Generation Sequencing).
Il test indaga le anomalie fetali sul sangue materno (mediante la NIPT, Non Invasive Prenatal Testing) e la ricerca sui genitori di mutazioni su geni specifici, associati a malattie genetiche trasmissibili al feto (Carrier Screening o Test del Portatore). Le malattie sono autosomico Recessive, presenza di una mutazione in entrambe le copie di un gene trasmesse da entrambi i genitori. Oppure X-Linked, caratteristica delle malattie che si manifestano nei maschi e per le quali le femmine sono portatrici sane.

FetalDNA - Complete Genetic Scan (CGS) si svolge dunque con l'applicazione di due esami diversi:
• FetalDNA Cariotipo + 21 Sindromi da Microdelezione + 8 Malattie Monogeniche
• Carrier Test Extended (o Test del Portatore Esteso)

L'esame, come per tutti i test non invasivi sul DNA Fetale, in caso di positività, può condurre al necessario riscontro con tecniche invasive (Villocentesi o Amniocentesi) per conferma o esclusione dei sospetti rilevati (rischio del 25% di malattia recessiva da genitori entrambi portatori sani, 50% di rischio di feti maschi affetti per malattia X-Linked per madre portatrice sana).
In tal caso si procederà, nel tempo più breve possibile, alla ricerca della patologia genetica nel feto mediante Diagnosi Prenatale Invasiva gratuita.

FETALDNA CARIOTIPO + 21 SINDROMI DA MICRODELEZIONE+ 8 MALATTIE MONOGENICHE

Il FetalDNA comprende il seguente studio:
1) **Lo screening del Cariotipo completo fetale.**
Accuratezza media del 99%.

2) **Lo screening di 21 sindromi da microdelezioni:**
Sindrome di Wolf-Hirschhorn, Sindrome HNPP, Sindrome di Jacobsen, Sindrome da delezione 18q, Sindrome da delezione 1p36, Sindrome di Alagille, Sindrome di Angelman, Sindrome di Rubinstein-Taybi, Sindrome di DiGeorge, Sindrome di WAGR, Sindrome di Cri-du-chat, Sindrome di Potocki-Shaffer, Sindrome di Langer-Giedion, Sindrome di Miller-Dieker, Sindrome di Smith-Magenis, Sindrome da delezione 1q21.1, Sindrome di Prader-Willi, Sindrome di Kleefstra, Sindrome di Williams, Sindrome di Phelan-Mcdermid, Sindrome di Koolen-de-Vries.
Accuratezza media, variabile in base alla frazione fetale e dal tipo di malattia, circa dell'85%

3) **Lo screening di 7 malattie monogeniche**, determinate da mutazioni spontanee ed associate a malattie Autosomiche Dominanti: Sindrome di Apert, Sindrome di Crouzon, Sindrome di Pfeiffer, Sindrome di Leopard, Sindrome di Noonan, Acondroplasia, Ipocondroplasia, Displasia tanatofora.
Accuratezza media del test, variabile in base alla frazione fetale, del 90%.

CARRIER TEST EXTENDED (o test del portatore esteso)

Test diagnostico eseguito sulla coppia di genitori.
Esso indica con certezza la presenza di mutazioni patologiche a carico della coppia, varianti queste trasmissibili al feto sia in modalità autosomico recessiva sia X-linked.
Anche in questo caso, per mutazioni presenti sulla coppia e per malattie genetiche importanti, si procede ad un esame di approfondimento mediante Amniocentesi o villocentesi allo scopo di verificare l'eventuale trasmissione al feto.

Altri approfondimenti potrebbero essere eseguiti sul DNA dei genitori o sui familiari diretti.

Il Carrier Test Extended o Test del Portatore comprende il seguente studio:
1) **Oltre 1300 malattie** genetiche Autosomiche Recessive o X-Linked trasmesse da entrambi i genitori portatori sani (nelle recessive) o dalla madre (nelle X-Linked). Le malattie sono sostenute da **più di 900 geni**.

Riferimenti bibliografici e linee guida sono riportati nel sito www.fetaldna.it