COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
CYP17A1	10q24.32	17,20-lyase deficiency, isolated	202110	🔴
CYP17A1	10q24.32	17-alpha-hydroxylase/17,20-lyase deficiency	202110	🔴
HSD17B10	Xp11.22	17-beta-hydroxysteroid dehydrogenase X deficiency	300438	🔴
HSD3B2	1p12	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810	🔴
HADH	4q25	3-hydroxyacyl-CoA dehydrogenase deficiency	231530	🔴
HIBCH	2q32.2	3-hydroxyisobutryl-CoA hydrolase deficiency	250620	🟡
HIBCH	2q32.2	3-hydroxyisobutryl-CoA hydrolase deficiency	250620	🔴
AUH	9q22.31	3-methylglutaconic aciduria, type I	250950	🔴
DNAJC19	3q26.33	3-methylglutaconic aciduria, type V	610198	🔴
NR5A1	9q33.3	46XY sex reversal 3	612965	🔴
FGD1	Xp11.22	Aarskog-Scott syndrome	305400	🟡
EDNRB	13q22.3	ABCD syndrome	600501	🔴
AAAS	12q13.13	Achalasia-addisonianism-alacrimia syndrome	231550	🔴
SLC26A2	5q32	Achondrogenesis Ib	600972	🔴
ACADM	1p31.1	Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	🔴
ADA	20q13.12	Adenosine deaminase deficiency, partial	102700	🔴
PKLR	1q22	Adenosine triphosphate, elevated, of erythrocytes	102900	🔴
CYP11B1	8q24.3	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	🔴
CYP21A2	6p21.33	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	201910	🟡
CYP11A1	15q24.1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743	🔴
NR5A1	9q33.3	Adrenocortical insufficiency		🔴
ABCD1	Xq28	Adrenoleukodystrophy	300100	🔴
ABCD1	Xq28	Adrenomyeloneuropathy, adult	300100	🔴
FGA	4q31.3	Afibrinogenemia, congenital	202400	🔴
BTK	Xq22.1	Agammaglobulinemia and isolated hormone deficiency	307200	🔴
BTK	Xq22.1	Agammaglobulinemia, X-linked 1	300755	🔴
SLC12A6	15q14	Agenesis of the corpus callosum with peripheral neuropathy	218000	🔴
TREX1	3p21.31	Aicardi-Goutieres syndrome 1, dominant and recessive	225750	🔴
RNASEH2B	13q14.3	Aicardi-Goutieres syndrome 2	610181	🔴
RNASEH2C	11q13.1	Aicardi-Goutieres syndrome 3	610329	🔴
RNASEH2A	19p13.2	Aicardi-Goutieres syndrome 4	610333	🔴
SAMHD1	20q11.23	Aicardi-Goutieres syndrome 5	612952	🔴
CYP11B1	8q24.3	Aldosteronism, glucocorticoid-remediable	103900	🔴
SLC16A2	Xq13.2	Allan-Herndon-Dudley syndrome	300523	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
RAG1	11p12	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion..	609889	🔴
ACAT1	11q22.3	Alpha-methylacetoacetic aciduria	203750	🔴
AMACR	5p13.2	Alpha-methylacyl-CoA racemase deficiency	614307	🟡
ATRX	Xq21.1	Alpha-thalassemia myelodysplasia syndrome, somatic	300448	🔴
ATRX	Xq21.1	Alpha-thalassemia/mental retardation syndrome	301040	🔴
COL4A5	Xq22.3	Alport syndrome	301050	🔴
COL4A3	2q36.3	Alport syndrome, autosomal dominant	104200	🔴
COL4A3	2q36.3	Alport syndrome, autosomal recessive	203780	🔴
COL4A	2q36.3	Alport syndrome, autosomal recessive	203780	🔴
ALMS1	2p13.1	Alstrom syndrome	203800	🔴
LAMB3	1q32.2	Amelogenesis imperfecta, type IA	104530	🔴
ST3GAL5	2p11.2	Amish infantile epilepsy syndrome	609056	🔴
FGA	4q31.3	Amyloidosis, familial visceral	105200	🔴
ALS2	2q33.1	Amyotrophic lateral sclerosis 2, juvenile	205100	🔴
RMRP	9p13.3	Anauxetic dysplasia	607095	🔴
AR	Xq12	Androgen insensitivity	300068	🔴
AR	Xq12	Androgen insensitivity, partial, with or without breast cancer	312300	🟡
MECP2	Xq28	Angelman syndrome	105830	🔴
UBE3A	15q11.2	Angelman syndrome	105830	🔴
UBE3A	15q11.2	Angelman syndrome	105830	🔴
CDKL5	Xp22.13	Angelman syndrome-like	105830	🔴
POR	7q11.23	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750	🔴
NBN	8q21.3	Aplastic anemia	609135	🔴
HSD11B2	16q22.1	Apparent mineralocorticoid excess	218030	🔴
ASL	7q11.21	Argininosuccinic aciduria	207900	🟡
DDC	7p12.1	Aromatic L-amino acid decarboxylase deficiency	608643	🔴
DSP	6p24.3	Arrhythmogenic right ventricular dysplasia 8	607450	🔴
ENPP1	6q23.2	Arterial calcification, generalized, of infancy, 1	208000	🔴
GLE1	9q34.11	Arthrogryposis, lethal, with anterior horn cell disease	611890	🔴
VPS33B	15q26.1	Arthrogryposis, renal dysfunction, and cholestasis 1	208085	🔴
VIPAR	14q24.3	Arthrogryposis, renal dysfunction, and cholestasis 2	613404	🔴
PRPS1	Xq22.3	Arts syndrome	301835	🔴
NLGN4X	Xp22.32-p22.31	Asperger syndrome susceptibility, X-linked 2	300497	🔴
TTPA	8q12.3	Ataxia with isolated vitamin E deficiency	277460	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
APTX	9p21.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	208920	🔴
ATM	11q22.3	Ataxia-telangiectasia	208900	🔴
SLC26A2	5q32	Atelosteogenesis II	256050	🔴
NLGN4X	Xp22.32-p22.31	Autism susceptibility, X-linked 2	300495	🔴
MECP2	Xq28	Autism susceptibility, X-linked 3	300496	🔴
RPL10	Xq28	Autism, susceptibility to, X-linked 5	300847	🔴
FAS	10q23.31	Autoimmune lymphoproliferative syndrome	601859	🟡
FAS	10q23.31	Autoimmune lymphoproliferative syndrome, type IA	601859	🟡
FASLG	1q24.3	Autoimmune lymphoproliferative syndrome, type IB	601859	🔴
AIRE	21q22.3	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300	🔴
PTEN	10q23.31	Bannayan-Riley-Ruvalcaba syndrome	153480	🟡
MKS1	17q22	Bardet-Biedl syndrome 13	615990	🔴
CEP290	12q21.32	Bardet-Biedl syndrome 14	615991	🔴
TMEM67	8q22.1	Bardet-Biedl syndrome 14, modifier of	209900	🔴
TAZ	Xq28	Barth syndrome	302060	🟡
GJB2	13q12.11	Bart-Pumphrey syndrome	149200	🔴
SLC12A1	15q21.1	Bartter syndrome, type 1	601678	🔴
KCNJ1	11q24.3	Bartter syndrome, type 2	241200	🔴
DMD	Xp21.2-p21.1	Becker muscular dystrophy	300376	🔴
NSD1	5q35.2-q35.3	Beckwith-Wiedemann syndrome	130650	🔴
COL6A1	21q22.3	Bethlem myopathy	158810	🔴
COL6A2	21q22.3	Bethlem myopathy	158810	🔴
COL6A3	2q37.3	Bethlem myopathy	158810	🔴
AMACR	5p13.2	Bile acid synthesis defect, congenital, 4	214950	🔴
BTD	3p25.1	Biotinidase deficiency	253260	🔴
BCS1L	2q35	Bjornstad syndrome	262000	🟡
BLM	15q26.1	Bloom syndrome	210900	🔴
COL1A1	17q21.33	Bone mineral density variation QTL, osteoporosis	166710	🔴
ZNF469	16q24.2	Brittle cornea syndrome 1	229200	🔴
SCNN1B	16p12.2	Bronchiectasis with or without elevated sweat chloride 1	211400	🔴
SCNN1A	12p13.31	Bronchiectasis with or without elevated sweat chloride 2	613021	🔴
SCNN1G	16p12.2	Bronchiectasis with or without elevated sweat chloride 3	613071	🔴
COL1A1	17q21.33	Caffey disease	114000	🟡

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
ASPA	17p13.2	Canavan disease	271900	●
CPS1	2q34	Carbamoylphosphate synthetase I deficiency	237300	●
SCO2	22q13.33	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	●
COX15	10q24.2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119	●
LMNA	1q22	Cardiomyopathy, dilated, 1A	115200	●
FKTN	9q31.2	Cardiomyopathy, dilated, 1X	611615	●
DMD	Xp21.2-p21.1	Cardiomyopathy, dilated, 3B	302045	●
DSP	6p24.3	Cardiomyopathy, dilated, with woolly hair and keratoderma	605676	●
SLC22A5	5q31.1	Carnitine deficiency, systemic primary	212140	●
SLC25A20	3p21.31	Carnitine-acylcarnitine translocase deficiency	212138	●
RAB23	6p11.2	Carpenter syndrome	201000	●
RMRP	9p13.3	Cartilage-hair hypoplasia	250250	●
NHS	Xp22.13	Cataract 40, X-linked	302200	●
EDN3	20q13.32	Central hypoventilation syndrome, congenital	209880	●
VLDLR	9p24.2	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	224050	●
SLC6A8	Xq28	Cerebral creatine deficiency syndrome 1	300352	●
GAMT	19p13.3	Cerebral creatine deficiency syndrome 2	612736	●
SNAP29	22q11.21	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma	609528	●
ERCC6	10q11.23	Cerebrooculofacioskeletal syndrome 1	214150	●
ERCC2	19q13.32	Cerebrooculofacioskeletal syndrome 2	610756	●
CYP27A1	2q35	Cerebrotendinous xanthomatosis	213700	●
PPT1	1p34.2	Ceroid lipofuscinosis, neuronal, 1	256730	●
CTSD	11p15.5	Ceroid lipofuscinosis, neuronal, 10	610127	●
TPP1	11p15.4	Ceroid lipofuscinosis, neuronal, 2	204500	●
CLN3	16p11.2	Ceroid lipofuscinosis, neuronal, 3	204200	●
CLN5	13q22.3	Ceroid lipofuscinosis, neuronal, 5	256731	●
CLN6	15q23	Ceroid lipofuscinosis, neuronal, 6	601780	●
MFSD8	4q28.2	Ceroid lipofuscinosis, neuronal, 7	610951	●
CLN8	8p23.3	Ceroid lipofuscinosis, neuronal, 8	600143	●
CLN8	8p23.3	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	610003	●
CLN6	15q23	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset	204300	●
GDAP1	8q21.11	Charcot-Marie-Tooth disease, axonal, type 2K	607831	●
IGHMBP2	11q13.3	Charcot-Marie-Tooth disease, axonal, type 2S	616155	●
GDAP1	8q21.11	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis	607706	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
MPZ	1q23.3	Charcot-Marie-Tooth disease, dominant intermediate D	607791	
PLEKHG5	1p36.31	Charcot-Marie-Tooth disease, recessive intermediate C	615376	
GDAP1	8q21.11	Charcot-Marie-Tooth disease, recessive intermediate, A	608340	
PMP22	17p12	Charcot-Marie-Tooth disease, type 1A	118220	
MPZ	1q23.3	Charcot-Marie-Tooth disease, type 1B	118200	
EGR2	10q21.3	Charcot-Marie-Tooth disease, type 1D	607678	
PMP22	17p12	Charcot-Marie-Tooth disease, type 1E	118300	
LMNA	1q22	Charcot-Marie-Tooth disease, type 2B1	605588	
MPZ	1q23.3	Charcot-Marie-Tooth disease, type 2I	607677	
MPZ	1q23.3	Charcot-Marie-Tooth disease, type 2J	607736	
GDAP1	8q21.11	Charcot-Marie-Tooth disease, type 4A	214400	
PRX	19q13.2	Charcot-Marie-Tooth disease, type 4F	614895	
FGD4	12p11.21	Charcot-Marie-Tooth disease, type 4H	609311	
PRPS1	Xq22.3	Charcot-Marie-Tooth disease, X-linked recessive, 5	311070	
LYST	1q42.3	Chediak-Higashi syndrome	214500	
TREX1	3p21.31	Chilblain lupus	610448	
SAMHD1	20q11.23	Chilblain lupus 2	614415	
ATP8B1	18q21.31	Cholestasis, benign recurrent intrahepatic	243300	
ABCB11	2q31.1	Cholestasis, benign recurrent intrahepatic, 2	605479	
ATP8B1	18q21.31	Cholestasis, intrahepatic, of pregnancy, 1	147480	
ABCB4	7q21.12	Cholestasis, intrahepatic, of pregnancy, 3	614972	
ATP8B1	18q21.31	Cholestasis, progressive familial intrahepatic 1	211600	
ABCB11	2q31.1	Cholestasis, progressive familial intrahepatic 2	601847	
ABCB4	7q21.12	Cholestasis, progressive familial intrahepatic 3	602347	
ARSE	Xp22.33	Chondrodysplasia punctata, X-linked recessive	302950	
PTH1R	3p21.31	Chondrodysplasia, Blomstrand type	215045	
ASS1	9q34.11	Citrullinemia	215700	
RPGRIP1L	16q12.2	COACH syndrome	216360	
TMEM67	8q22.1	COACH syndrome	216360	
ERCC8	5q12.1	Cockayne syndrome, type A	216400	
ERCC6	10q11.23	Cockayne syndrome, type B	133540	
COQ2	4q21.23	Coenzyme Q10 deficiency, primary, 1	607426	
PDSS1	10p12.1	Coenzyme Q10 deficiency, primary, 2	614651	
PDSS2	6q21	Coenzyme Q10 deficiency, primary, 3	614652	
ADCK3	1q42.13	Coenzyme Q10 deficiency, primary, 4	612016	
COQ9	16q21	Coenzyme Q10 deficiency, primary, 5	614654	
RPS6KA3	Xp22.12	Coffin-Lowry syndrome	303600	
VPS13B	8q22.2	Cohen syndrome	216550	
CRLF1	19p13.11	Cold-induced sweating syndrome 1	272430	
ENPP1	6q23.2	Cole disease	615522	

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
RAG1	11p12	Combined cellular and humoral immune defects with granulomas	233650	●
RAG2	11p12	Combined cellular and humoral immune defects with granulomas	233650	●
IL2R	Xq13.1	Combined immunodeficiency, X-linked, moderate	312863	●
GFM1	3q25.32	Combined oxidative phosphorylation deficiency 1	609060	●
MRPS16	10q22.2	Combined oxidative phosphorylation deficiency 2	610498	●
TSFM	12q14.1	Combined oxidative phosphorylation deficiency 3	610505	●
TUFM	16p11.2	Combined oxidative phosphorylation deficiency 4	610678	●
MRPS22	3q23	Combined oxidative phosphorylation deficiency 5	611719	●
PSAP	10q22.1	Combined SAP deficiency	611721	●
NDUFS6	5p15.33	Complex I, mitochondrial respiratory chain, deficiency of	252010	●
PMM2	16p13.2	Congenital disorder of glycosylation, type Ia	212065	●
MPI	15q24.1	Congenital disorder of glycosylation, type Ib	602579	●
ALG6	1p31.3	Congenital disorder of glycosylation, type Ic	603147	●
ALG3	3q27.1	Congenital disorder of glycosylation, type Id	601110	●
DPM1	20q13.13	Congenital disorder of glycosylation, type Ie	608799	●
MPDU1	17p13.1	Congenital disorder of glycosylation, type If	609180	●
ALG12	22q13.33	Congenital disorder of glycosylation, type Ig	607143	●
ALG8	11q14.1	Congenital disorder of glycosylation, type Ih	608104	●
ALG2	9q22.33	Congenital disorder of glycosylation, type Ii	607906	●
MGAT2	14q21.3	Congenital disorder of glycosylation, type IIa	212066	●
MOGS	2p13.1	Congenital disorder of glycosylation, type IIb	606056	●
SLC35C1	11p11.2	Congenital disorder of glycosylation, type IIc	266265	●
B4GALT1	9p21.1	Congenital disorder of glycosylation, type IId	607091	●
COG7	16p12.2	Congenital disorder of glycosylation, type IIe	608779	●
SLC35A1	6q15	Congenital disorder of glycosylation, type IIf	603585	●
COG1	17q25.1	Congenital disorder of glycosylation, type IIg	611209	●
COG8	16q22.1	Congenital disorder of glycosylation, type IIh	611182	●
DPAGT1	11q23.3	Congenital disorder of glycosylation, type Ij	608093	●
ALG1	16p13.3	Congenital disorder of glycosylation, type Ik		●
ALG9	11q23.1	Congenital disorder of glycosylation, type Il	608776	●
DOLK	9q34.11	Congenital disorder of glycosylation, type Im	610768	●
RFT1	3p21.1	Congenital disorder of glycosylation, type In	612015	●
SRD5A3	4q12	Congenital disorder of glycosylation, type Iq	612379	●
ZIC3	Xq26.3	Congenital heart defects, nonsyndromic, 1, X-linked	306955	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
SLC4A11	20p13	Corneal dystrophy, Fuchs endothelial, 4	613268	●
SLC4A11	20p13	Corneal endothelial dystrophy 2, autosomal recessive	217700	●
SLC4A11	20p13	Corneal endothelial dystrophy and perceptive deafness	217400	●
L1CAM	Xq28	Corpus callosum, partial agenesis of	304100	●
PTEN	10q23.31	Cowden syndrome 1	158350	●
CPT1A	11q13.3	CPT deficiency, hepatic, type IA	255120	●
CPT2	1p32.3	CPT deficiency, hepatic, type II	600649	●
CPT2	1p32.3	CPT II deficiency, lethal neonatal	608836	●
EFNB1	Xq13.1	Craniofrontonasal dysplasia	304110	●
L1CAM	Xq28	CRASH syndrome	303350	●
ATR	3q23	Cutaneous telangiectasia and cancer syndrome, familial	614564	●
FBLN5	14q32.12	Cutis laxa, autosomal recessive, type IA	219100	●
EFEMP2	11q13.1	Cutis laxa, autosomal recessive, type IB	614437	●
ATP6V0A2	12q24.31	Cutis laxa, autosomal recessive, type IIA	219200	●
CFTR	7q31.2	Cystic fibrosis	219700	●
CTNS	17p13.2	Cystinosis, atypical nephropathic	219800	●
CTNS	17p13.2	Cystinosis, late-onset juvenile or adolescent nephropathic	219900	●
CTNS	17p13.2	Cystinosis, nephropathic	219800	●
CTNS	17p13.2	Cystinosis, ocular nonnephropathic	219750	●
HSD17B4	5q23.1	D-bifunctional protein deficiency	261515	●
SLC26A2	5q32	De la Chapelle dysplasia	256050	●
ERCC6	10q11.23	De Sanctis-Cacchione syndrome	278800	●
MYO7A	11q13.5	Deafness, autosomal dominant 11	601317	●
GJB2	13q12.11	Deafness, autosomal dominant 3A	601544	●
CDH23	10q22.1	Deafness, autosomal recessive 12	601386	●
USH1C	11p15.1	Deafness, autosomal recessive 18A	602092	●
GJB2	13q12.11	Deafness, autosomal recessive 1A	220290	●
MYO7A	11q13.5	Deafness, autosomal recessive 2	600060	●
PRPS1	Xq22.3	Deafness, X-linked 1	304500	●
TIMM8A	Xq22.1	Deafness, X-linked 1, progressive	300066	●
F9	Xq27.1	Deep venous thrombosis, protection against	300807	●
EGR2	10q21.3	Dejerine-Sottas disease	145900	●
MPZ	1q23.3	Dejerine-Sottas disease	145900	●
PMP22	17p12	Dejerine-Sottas disease	145900	●
PRX	19q13.2	Dejerine-Sottas disease	145900	●
HBB	11p15.4	Delta-beta thalassemia	141749	●
CLCN5	Xp11.23-p11.22	Dent disease	300009	●
OCRL	Xq25-q26	Dent disease 2	300555	●
DHCR24	1p32.3	Desmosterolosis	602398	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
INSR	19p13.2	Diabetes mellitus, insulin-resistant, with acanthosis nigricans	610549	🟡
ABCC8	11p15.1	Diabetes mellitus, noninsulin-dependent	125853	🟡
ENPP1	6q23.2	Diabetes mellitus, non-insulin-dependent, susceptibility to	125853	🟡
ABCC8	11p15.1	Diabetes mellitus, permanent neonatal	606176	🔴
ABCC8	11p15.1	Diabetes mellitus, transient neonatal 2	610374	🔴
FOXP3	Xp11.23	Diabetes mellitus, type I, susceptibility to	222100	🔴
NEUROG3	10q22.1	Diarrhea 4, malabsorptive, congenital	610370	🔴
SLC26A2	5q32	Diastrophic dysplasia	222600	🔴
SLC26A2	5q32	Diastrophic dysplasia, broad bone-platyspondylic variant	222600	🔴
DLD	31.1	Dihydrolipoamide dehydrogenase deficiency	246900	🔴
DPYD	1p21.3	Dihydropyrimidine dehydrogenase deficiency	274270	🔴
DSP	6p24.3	Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	615821	🔴
POR	7q11.23	Disordered steroidogenesis due to cytochrome P450 oxidoreductase	613571	🔴
LRP2	2q31.1	Donnai-Barrow syndrome	222448	🔴
DMD	Xp21.2-p21.1	Duchenne muscular dystrophy	310200	🔴
G6PC3	17q21.31	Dursun syndrome	612541	🔴
IKBKAP	9q31.3	Dysautonomia, familial	223900	🔴
FGA	4q31.3	Dysfibrinogenemia, congenital	616004	🟡
DKC1	Xq28	Dyskeratosis congenita, X-linked	305000	🔴
PLG	6q26	Dysplasminogenemia	217090	🔴
HSPG2	1p36.12	Dyssegmental dysplasia, Silverman-Handmaker type	224410	🔴
COL6A3	2q37.3	Dystonia 27	616411	🔴
COL7A1	3p21.31	EBD inversa	226600	🔴
COL7A1	3p21.31	EBD, Bart type	132000	🔴
COL7A1	3p21.31	EBD, localisata variant		🔴
EDA	Xq13.1	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	🔴
IKBKG	Xq28	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291	🔴
IKBKG	Xq28	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301	🔴
COL1A2	7q21.3	Ehlers-Danlos syndrome, cardiac valvular form	225320	🔴
COL1A1	17q21.33	Ehlers-Danlos syndrome, classic	130000	🔴
PLOD1	1p36.22	Ehlers-Danlos syndrome, type VI	225400	🔴
COL1A1	17q21.33	Ehlers-Danlos syndrome, type VIIA	130060	🔴
COL1A2	7q21.3	Ehlers-Danlos syndrome, type VIIB	130060	🔴
PTH1R	3p21.31	Eiken syndrome	600002	🔴
EVC	4p16.2	Ellis-van Creveld syndrome	225500	🔴
EVC2	4p16.2	Ellis-van Creveld syndrome	225500	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
LMNA	1q22	Emery-Dreifuss muscular dystrophy 3, AR	181350	●
MECP2	Xq28	Encephalopathy, neonatal severe	300673	●
COL7A1	3p21.31	Epidermolysis bullosa dystrophica, AR	226600	●
ITGB4	17q25.1	Epidermolysis bullosa of hands and feet	131800	●
COL7A1	3p21.31	Epidermolysis bullosa pruriginosa	604129	●
PLEC	8q24.3	Epidermolysis bullosa simplex with pyloric atresia	612138	●
PLEC	8q24.3	Epidermolysis bullosa simplex, Ogna type	131950	●
LAMA3	18q11.2	Epidermolysis bullosa, generalized atrophic benign	226650	●
LAMA3	18q11.2	Epidermolysis bullosa, junctional, Herlitz type	226700	●
LAMB3	1q32.2	Epidermolysis bullosa, junctional, Herlitz type	226700	●
LAMC2	1q25.3	Epidermolysis bullosa, junctional, Herlitz type	226700	●
COL17A1	10q24.3-q25.1	Epidermolysis bullosa, junctional, non-Herlitz type	226650	●
ITGB4	17q25.1	Epidermolysis bullosa, junctional, non-Herlitz type	226650	●
LAMB3	1q32.2	Epidermolysis bullosa, junctional, non-Herlitz type	226650	●
LAMC2	1q25.3	Epidermolysis bullosa, junctional, non-Herlitz type	226650	●
ITGB4	17q25.1	Epidermolysis bullosa, junctional, with pyloric atresia	226730	●
ITGA6	2q31.1	Epidermolysis bullosa, junctional, with pyloric stenosis	226730	●
DSP	6p24.3	Epidermolysis bullosa, lethal acantholytic	609638	●
COL7A1	3p21.31	Epidermolysis bullosa, pretibial	131850	●
RELN	7q22.1	Epilepsy, familial temporal lobe, 7	616436	●
CSTB	21q22.3	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	254800	●
EPM2A	6q24.3	Epilepsy, progressive myoclonic 2A (Lafora)	254780	●
NHLRC1	6p22.3	Epilepsy, progressive myoclonic 2B (Lafora)	254780	●
ALDH7A1	5q23.2	Epilepsy, pyridoxine-dependent	266100	●
ARX	Xp21.3	Epileptic encephalopathy, early infantile, 1	308350	●
ST3GAL3	1p34.1	Epileptic encephalopathy, early infantile, 15	615006	●
CDKL5	Xp22.13	Epileptic encephalopathy, early infantile, 2	300672	●
SLC25A22	11p15.5	Epileptic encephalopathy, early infantile, 3	609304	●
ARHGEF9	Xq11.1-q11.2	Epileptic encephalopathy, early infantile, 8	300607	●
PCDH19	Xq22.1	Epileptic encephalopathy, early infantile, 9	300088	●
SLC26A2	5q32	Epiphyseal dysplasia, multiple, 4	226900	●
HBA1	16p13.3	Thalassemia, alpha-	141750	●
HBB	11p15.4	Thalassemia, beta-	613985	●
CHRNG	2q37.1	Escobar syndrome	265000	●
ETHE1	19q13.31	Ethylmalonic encephalopathy	602473	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
NDP	Xp11.3	Exudative vitreoretinopathy 2, X-linked	305390	🟡
GLA	Xq22.1	Fabry disease	301500	🔴
PTH1R	3p21.31	Failure of tooth eruption, primary	125350	🔴
MEFV	16p13.3	Familial Mediterranean fever, AR	249100	🔴
FANCC	9q22.32	Fanconi anemia, complementation group C	227645	🔴
ERCC4	16p13.12	Fanconi anemia, complementation group Q	610965	🔴
HADHA	2p23.3	Fatty liver, acute, of pregnancy	609016	🔴
G6PD	Xq28	Favism	134700	🟡
GPR98	5q14.3	Febrile seizures, familial, 4	604352	🔴
RAPSN	11p11.2	Fetal akinesia deformation sequence	208150	🔴
DOK7	4p16.3	Fetal akinesia deformation sequence	208150	🔴
CASK	Xp11.4	FG syndrome 4	300422	🔴
FRAS1	4q21.21	Fraser syndrome	219000	🔴
FREM2	13q13.3	Fraser syndrome	219000	🔴
ALDOB	9q31.1	Fructose intolerance	229600	🟡
FUCA1	1p36.11	Fucosidosis	230000	🔴
WNT7A	3p25.1	Fuhrmann syndrome	228930	🔴
GALK1	17q25.1	Galactokinase deficiency with cataracts	230200	🟡
GALT	9p13.3	Galactosemia	230400	🔴
ABCB4	7q21.12	Gallbladder disease 1	600803	🔴
PSAP	10q22.1	Gaucher disease, atypical	610539	🔴
GBA	1q22	Gaucher disease, perinatal lethal	608013	🔴
GBA	1q22	Gaucher disease, type I	230800	🔴
GBA	1q22	Gaucher disease, type II	230900	🔴
GBA	1q22	Gaucher disease, type III	231000	🔴
GBA	1q22	Gaucher disease, type IIIC	231005	🔴
ADAMTSL2	9q34.2	Geleophysic dysplasia 1	231050	🔴
ETFA	15q24.2-q24.3	Glutaric acidemia IIA	231680	🔴
ETFB	19q13.41	Glutaric acidemia IIB	231680	🔴
ETFDH	4q32.1	Glutaric acidemia IIC	231680	🔴
GCDH	19p13.2	Glutaricaciduria, type I	231670	🔴
GSS	20q11.22	Glutathione synthetase deficiency	266130	🔴
AMT	3p21.31	Glycine encephalopathy	605899	🔴
GCSH	16q23.2	Glycine encephalopathy	605899	🔴
GLDC	9p24.1	Glycine encephalopathy	605899	🔴
G6PC	17q21.31	Glycogen storage disease Ia	232200	🔴
SLC37A4	11q23.3	Glycogen storage disease Ib	232220	🔴
SLC37A4	11q23.3	Glycogen storage disease Ic	232240	🔴
GAA	17q25.3	Glycogen storage disease II	232300	🔴
AGL	1p21.2	Glycogen storage disease IIIa	232400	🔴
AGL	1p21.2	Glycogen storage disease IIIb	232400	🔴
GBE1	3p12.2	Glycogen storage disease IV	232500	🔴
GLB1	3p22.3	GM1-gangliosidosis, type I	230500	🔴
GLB1	3p22.3	GM1-gangliosidosis, type II	230600	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
GLB1	3p22.3	GM1-gangliosidosis, type III	230650	🔴
HEXA	15q23	GM2-gangliosidosis, several forms	272800	🔴
PRPS1	Xq22.3	Gout, PRPS-related	300661	🔴
BCS1L	2q35	GRACILE syndrome	603358	🔴
LBR	1q42.12	Greenberg skeletal dysplasia	215140	🔴
MYO5A	15q21.2	Griscelli syndrome, type 1	214450	🔴
RAB27A	15q21.3	Griscelli syndrome, type 2	607624	🔴
HESX1	3p14.3	Growth hormone deficiency with pituitary anomalies	182230	🔴
PANK2	20p13	HARP syndrome	607236	🔴
HP	12q24.31	Hawkinsinuria	140350	🟡
HADHA	2p23.3	HELLP syndrome, maternal, of pregnancy	609016	🔴
COL4A3	2q36.3	Hematuria, benign familial	141200	🔴
COL4A	2q36.3	Hematuria, familial benign		🔴
HFE2	1q21.1	Hemochromatosis, type 2A	602390	🔴
HAMP	19q13.12	Hemochromatosis, type 2B	613313	🔴
HBA1	16p13.3	Hemoglobin H disease, nondeletional	613978	🔴
G6PD	Xq28	Hemolytic anemia due to G6PD deficiency	300908	🔴
GSS	20q11.22	Hemolytic anemia due to glutathione synthetase deficiency	231900	🔴
PRF1	10q22.1	Hemophagocytic lymphohistiocytosis, familial, 2	603553	🔴
UNC13D	17q25.1	Hemophagocytic lymphohistiocytosis, familial, 3	608898	🔴
STX11	6q24.2	Hemophagocytic lymphohistiocytosis, familial, 4	603552	🔴
STXBP2	19p13.2	Hemophagocytic lymphohistiocytosis, familial, 5	613101	🔴
F8	Xq28	Hemophilia A	306700	🔴
F9	Xq27.1	Hemophilia B	306900	🔴
SCO1	17p13.1	Hepatic failure, early onset, and neurologic disorder	603644	🔴
SP110	2q37.1	Hepatic venoocclusive disease with immunodeficiency	235550	🔴
HBB	11p15.4	Hereditary persistence of fetal hemoglobin	141749	🔴
AP3B1	5q14.1	Hermansky-Pudlak syndrome 2	608233	🔴
PLDN	15q21.1	Hermansky-pudlak syndrome 9	614171	🔴
UNC93B1	11q13.2	Herpes simplex encephalitis, susceptibility to, 1	610551	🔴
TLR3	4q35.1	Herpes simplex encephalitis, susceptibility to, 2	613002	🔴
ZIC3	Xq26.3	Heterotaxy, visceral, 1, X-linked	306955	🔴
HEXA	15q23	Hex A pseudodeficiency	272800	🔴
TLR3	4q35.1	HIV1 infection, resistance to	609423	🔴
HMGCL	1p36.11	HMG-CoA lyase deficiency	246450	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
HLCS	21q22.13	Holocarboxylase synthetase deficiency	253270	🔴
CBS	21q22.3	Homocystinuria, B6-responsive and nonresponsive types	236200	🔴
LMNA	1q22	Hutchinson-Gilford progeria	176670	🔴
ANTXR2	4q21.21	Hyaline fibromatosis syndrome	228600	🔴
ARX	Xp21.3	Hydranencephaly with abnormal genitalia	300215	🔴
L1CAM	Xq28	Hydrocephalus due to aqueductal stenosis	307000	🔴
L1CAM	Xq28	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction	307000	🔴
L1CAM	Xq28	Hydrocephalus with Hirschsprung disease	307000	🔴
CYP21A2	6p21.33	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	201910	🔴
FH	19p13.2	Hypercholesterolemia, familial	143890	🟡
MVK	12q24.11	Hyper-IgD syndrome	260920	🔴
DOCK8	9p24.3	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	🔴
ABCC8	11p15.1	Hyperinsulinemic hypoglycemia, familial, 1	256450	🔴
HADH	4q25	Hyperinsulinemic hypoglycemia, familial, 4	609975	🔴
INSR	19p13.2	Hyperinsulinemic hypoglycemia, familial, 5	609968	🔴
SLC25A15	13q14.11	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970	🔴
FGA	4q31.3	Hypodysfibrinogenemia, congenital	616004	🔴
ABCC8	11p15.1	Hypoglycemia of infancy, leucine-sensitive	240800	🔴
GNRHR	4q13.2	Hypogonadotropic hypogonadism 7 without anosmia	146110	🔴
CLDN19	1p34.2	Hypomagnesemia 5, renal, with ocular involvement	248190	🔴
TBCE	1q42.3	Hypoparathyroidism-retardation-dysmorphism syndrome	241410	🔴
ALPL	1p36.12	Hypophosphatasia, childhood	241510	🔴
ALPL	1p36.12	Hypophosphatasia, infantile	241500	🔴
CLCN5	Xp11.23-p11.22	Hypophosphatemic rickets	300554	🟡
DMP1	4q22.1	Hypophosphatemic rickets, AR	241520	🔴
ENPP1	6q23.2	Hypophosphatemic rickets, autosomal recessive, 2	613312	🔴
AR	Xq12	Hypospadias 1, X-linked	300633	🔴
TSHB	1p13.2	Hypothryoidism, congenital, nongoitrous 4	275100	🔴
GJB2	13q12.11	Hystrix-like ichthyosis with deafness	602540	🔴
ABCA12	2q35	Ichthyosis, autosomal recessive 4B (harlequin)	242500	🔴
TGM1	14q12	Ichthyosis, congenital, autosomal recessive 1	242300	🔴
CLDN1	3q28	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	607626	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
MBTPS2	Xp22.12-p22.11	IFAP syndrome with or without BRESHECK syndrome	308205	🔴
STIM1	11p15.4	Immunodeficiency 10	612783	🔴
CD3G	11q23.3	Immunodeficiency 17, CD3 gamma deficient	615607	🔴
CD3E	11q23.3	Immunodeficiency 18	615615	🔴
CD3E	11q23.3	Immunodeficiency 18, SCID variant	615615	🔴
CD3D	11q23.3	Immunodeficiency 19	615617	🔴
CD247	1q24.2	Immunodeficiency 25	610163	🔴
IFNGR1	6q23.3	Immunodeficiency 27A, mycobacteriosis, AR	209950	🔴
IFNGR1	6q23.3	Immunodeficiency 27B, mycobacteriosis, AD	615978	🔴
IFNGR2	21q22.11	Immunodeficiency 28, mycobacteriosis	614889	🔴
IL12B	5q33.3	Immunodeficiency 29, mycobacteriosis	614890	🔴
IL12RB1	19p13.11	Immunodeficiency 30	614891	🔴
STAT1	2q32.2	Immunodeficiency 31B, mycobacterial and viral infections, autosomal reces.	613796	🔴
IKBKG	Xq28	Immunodeficiency 33	300636	🔴
TYK2	19p13.2	Immunodeficiency 35	611521	🔴
ORAI1	12q24.31	Immunodeficiency 9	612782	🔴
ICOS	2q33.2	Immunodeficiency, common variable, 1	607594	🔴
CD19	16p11.2	Immunodeficiency, common variable, 3	613493	🔴
IKBKG	Xq28	Immunodeficiency, isolated	300584	🔴
CD40LG	Xq26.3	Immunodeficiency, X-linked, with hyper-IgM	308230	🔴
DNMT3B	20q11.21	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	🔴
FOXP3	Xp11.23	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790	🔴
IKBKG	Xq28	Incontinentia pigmenti	308300	🔴
PLA2G6	22q13.1	Infantile neuroaxonal dystrophy 1	256600	🔴
NTRK1	1q23.1	Insensitivity to pain, congenital, with anhidrosis	256800	🔴
IL1RN	2q13	Interleukin 1 receptor antagonist deficiency	612852	🔴
IVD	15q15.1	Isovaleric acidemia	243500	🔴
TIMM8A	Xq22.1	Jensen syndrome	311150	🔴
UBR1	15q15.2	Johanson-Blizzard syndrome	243800	🔴
OFD1	Xp22.2	Joubert syndrome 10	300804	🔴
NPHP1	2q13	Joubert syndrome 4	609583	🔴
CEP290	12q21.32	Joubert syndrome 5	610188	🔴
TMEM67	8q22.1	Joubert syndrome 6	610688	🔴
RPGRIP1L	16q12.2	Joubert syndrome 7	611560	🔴
AHI1	6q23.3	Joubert syndrome-3	608629	🔴
SRD5A3	4q12	Kahrizi syndrome	612713	🔴
HPRT	Xq26.2-q26.3	KELLEY-SEEGMILLER SYNDROME	300323	🟡
TBCE	1q42.3	Kenny-Caffey syndrome-1	244460	🔴
GJB2	13q12.11	Keratitis-ichthyosis-deafness syndrome	148210	🔴
GJB2	13q12.11	Keratoderma, palmoplantar, with deafness	148350	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
MBTPS2	Xp22.12-p22.11	Keratosis follicularis spinulosa decalvans, X-linked	308800	
DSP	6p24.3	Keratosis palmoplantaris striata II	612908	
GALC	14q31.3	Krabbe disease	245200	
PSAP	10q22.1	Krabbe disease, atypical	611722	
PDHX	11p13	Lacticacidemia due to PDX1 deficiency	245349	
LAMA3	18q11.2	Laryngoonychocutaneous syndrome	245660	
SC5DL	11q23.3	Lathosterolosis	607330	
HADHA	2p23.3	LCHAD deficiency	609016	
FH	19p13.2	LDL cholesterol level QTL2	143890	
CEP290	12q21.32	Leber congenital amaurosis 10	611755	
BCS1L	2q35	Leigh syndrome	256000	
NDUFAF	5q12.1	Leigh syndrome	256000	
NDUFS4	5q11.2	Leigh syndrome	256000	
NDUFS7	19p13.3	Leigh syndrome	256000	
COX15	10q24.2	Leigh syndrome due to cytochrome c oxidase deficiency	256000	
NDUFS3	11p11.2	Leigh syndrome due to mitochondrial complex I deficiency	256000	
NDUFS8	11q13.2	Leigh syndrome due to mitochondrial complex I deficiency	256000	
COX10	17p12	Leigh syndrome due to mitochondrial COX4 deficiency	256000	
SURF1	9q34.2	Leigh syndrome, due to COX deficiency	256000	
LRPPRC	2p21	Leigh syndrome, French-Canadian type	220111	
INSR	19p13.2	Leprechaunism	246200	
HPRT	Xq26.2-q26.3	Lesch-Nyhan syndrome	300322	
ERBB3	12q13.2	Lethal congenital contractural syndrome 2	607598	
GLE1	9q34.11	Lethal congenital contracture syndrome 1	253310	
NBN	8q21.3	Leukemia, acute lymphoblastic	613065	
NSD1	5q35.2-q35.3	Leukemia, acute myeloid	601626	
FERMT3	11q13.1	Leukocyte adhesion deficiency, type III	612840	
GJC2	1q42.13	Leukodystrophy, hypomyelinating, 2	608804	
FAM126A	7p15.3	Leukodystrophy, hypomyelinating, 5	610532	
GBA	1q22	Lewy body dementia, susceptibility to	127750	
LHCGR	2p16.3	Leydig cell adenoma, somatic, with precocious puberty	176410	
LHCGR	2p16.3	Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320	
LHCGR	2p16.3	Leydig cell hypoplasia with pseudohermaphroditism	238320	
PTEN	10q23.31	Lhermitte-Duclos syndrome	158350	
SCNN1B	16p12.2	Liddle syndrome	177200	

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
SCNN1G	16p12.2	Liddle syndrome	177200	
LIG4	13q33.3	LIG4 syndrome	606593	
LMNA	1q22	Lipodystrophy, familial partial, 2	151660	
STAR	8p11.23	Lipoid adrenal hyperplasia	201710	
RELN	7q22.1	Lissencephaly 2 (Norman-Roberts type)	257320	
TUBA1A	12q13.12	Lissencephaly 3	611603	
DCX	Xq23	Lissencephaly, X-linked	300067	
ARX	Xp21.3	Lissencephaly, X-linked 2	300215	
OCRL	Xq25-q26	Lowe syndrome	309000	
MED12	Xq13.1	Lujan-Fryns syndrome	309520	
LHCGR	2p16.3	Luteinizing hormone resistance, female	238320	
GJC2	1q42.13	Lymphedema, hereditary, IC	613480	
PRF1	10q22.1	Lymphoma, non-Hodgkin	605027	
SH2D1A	Xq25	Lymphoproliferative syndrome, X-linked, 1	308240	
XIAP	Xq25	Lymphoproliferative syndrome, X-linked, 2	300635	
PTEN	10q23.31	Macrocephaly/autism syndrome	605309	
MYD88	3p22.2	Macroglobulinemia, Waldenstrom, somatic	153600	
ERCC6	10q11.23	Macular degeneration, age-related, susceptibility to 5	613761	
MFSD8	4q28.2	Macular dystrophy with central cone involvement	616170	
LMNA	1q22	Malouf syndrome	212112	
LMNA	1q22	Mandibuloacral dysplasia	248370	
ZMPSTE24	1p34.2	Mandibuloacral dysplasia with type B lipodystrophy	608612	
MAN2B1	19p13.2	Mannosidosis, alpha-, types I and II	248500	
BCKDHA	19q13.2	Maple syrup urine disease, type Ia	248600	
BCKDHB	6q14.1	Maple syrup urine disease, type Ib	248600	
DBT	1p21.2	Maple syrup urine disease, type II	248600	
SIL1	5q31.2	Marinesco-Sjogren syndrome	248800	
RAB3GAP2	1q41	Martsolf syndrome	212720	
L1CAM	Xq28	MASA syndrome	303350	
PYGM	11q13.1	McArdle disease	232600	
MKS1	17q22	Meckel syndrome 1	249000	
TMEM67	8q22.1	Meckel syndrome 3	607361	
CEP290	12q21.32	Meckel syndrome 4	611134	
RPGRIP1L	16q12.2	Meckel syndrome 5	611561	
NPHP3	3q22.1	Meckel syndrome 7	267010	
MLC1	22q13.33	Megalencephalic leukoencephalopathy with subcortical cysts	604004	
ATP7A	Xq21.1	Menkes disease	309400	
CASK	Xp11.4	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749	
DOCK8	9p24.3	Mental retardation, autosomal dominant 2	614113	
PRSS12	4q26	Mental retardation, autosomal recessive 1	249500	

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
ST3GAL3	1p34.1	Mental retardation, autosomal recessive 12	611090	●
TRAPPC9	8q24.3	Mental retardation, autosomal recessive 13	613192	●
NSUN2	5p15.31	Mental retardation, autosomal recessive 5	611091	●
TUSC3	8p22	Mental retardation, autosomal recessive 7	611093	●
GRIK2	6q16.3	Mental retardation, autosomal recessive, 6	611092	●
CASK	Xp11.4	Mental retardation, with or without nystagmus	300422	●
NLGN4X	Xp22.32-p22.31	Mental retardation, X-linked	300495	●
RPS6KA3	Xp22.12	Mental retardation, X-linked 19	300844	●
IL1RAPL1	Xp21.3-p21.2	Mental retardation, X-linked 21/34	300143	●
ARX	Xp21.3	Mental retardation, X-linked 29 and others	300419	●
PAK3	Xq23	Mental retardation, X-linked 30/47	300558	●
GDI1	Xq28	Mental retardation, X-linked 41	300849	●
ARHGEF6	Xq26.3	Mental retardation, X-linked 46	300436	●
ACSL4	Xq23	Mental retardation, X-linked 63	300387	●
RAB39B	Xq28	Mental retardation, X-linked 72	300271	●
AGTR2	Xq24	Mental retardation, X-linked 88	300852	●
FTSJ1	Xp11.23	Mental retardation, X-linked 9	309549	●
DLG3	Xq13.1	Mental retardation, X-linked 90	300850	●
BRWD3	Xq21.1	Mental retardation, X-linked 93	300659	●
SYP	Xp11.23	Mental retardation, X-linked 96	300802	●
ZNF711	Xq21.1	Mental retardation, X-linked 97	300803	●
HSD17B10	Xp11.22	Mental retardation, X-linked syndromic 10	300220	●
FGD1	Xp11.22	Mental retardation, X-linked syndromic 16	305400	●
AP1S2	Xp22.2	Mental retardation, X-linked syndromic 5	304340	●
SLC9A6	Xq26.3	Mental retardation, X-linked syndromic, Christianson type	300243	●
MECP2	Xq28	Mental retardation, X-linked syndromic, Lubs type	300260	●
UBE2A	Xq24	Mental retardation, X-linked syndromic, Nascimento-type	300860	●
ZDHHC9	Xq26.1	Mental retardation, X-linked syndromic, Raymond type	300799	●
HUWE1	Xp11.22	Mental retardation, X-linked syndromic, Turner type	300706	●
AFF2 (FMR2)	Xq28	Mental retardation, X-linked, FRAXE type	309548	●
SMS	Xp22.11	Mental retardation, X-linked, Snyder-Robinson type	309583	●
MECP2	Xq28	Mental retardation, X-linked, syndromic 13	300055	●
UPF3B	Xq24	Mental retardation, X-linked, syndromic 14	300676	●
CUL4B	Xq24	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354	●
KDM5C	Xp11.22	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
OPHN1	Xq12	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486	●
SOX3	Xq27.1	Mental retardation, X-linked, with isolated growth hormone deficiency	300123	●
ATRX	Xq21.1	Mental retardation-hypotonic facies syndrome, X-linked	309580	●
ARSA	22q13.33	Metachromatic leukodystrophy	250100	●
PSAP	10q22.1	Metachromatic leukodystrophy due to SAP-b deficiency	249900	●
PTH1R	3p21.31	Metaphyseal chondrodysplasia, Murk Jansen type	156400	●
RMRP	9p13.3	Metaphyseal dysplasia without hypotrichosis	250460	●
MMACHC	1p34.1	Methylmalonic aciduria and homocystinuria, cblC type	277400	●
MUT	6p12.3	Methylmalonic aciduria, mut(0) type	251000	●
MMAA	4q31.21	Methylmalonic aciduria, vitamin B12-responsive	251100	●
MMAB	12q24.11	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	●
MVK	12q24.11	Mevalonic aciduria	610377	●
STRA6	15q24.1	Microphthalmia, isolated, with coloboma 8	601186	●
BCOR	Xp11.4	Microphthalmia, syndromic 2	300166	●
STRA6	15q24.1	Microphthalmia, syndromic 9	601186	●
IL1RN	2q13	Microvascular complications of diabetes 4	612628	●
NDUFA7	19p13.3	Mitochondrial Complex 1 Deficiency (MT-C1D)	252010	●
NDUFA1	Xq24	Mitochondrial complex I deficiency	252010	●
NDUFAF4	6q16.1	Mitochondrial complex I deficiency	252010	●
NDUFS3	11p11.2	Mitochondrial complex I deficiency	252010	●
NDUFS4	5q11.2	Mitochondrial complex I deficiency	252010	●
NDUFS5	1p34.3	Mitochondrial complex I deficiency	252010	●
NDUFV1	11q13.2	Mitochondrial complex I deficiency	252010	●
ACAD9	3q21.3	Mitochondrial complex I deficiency due to ACAD9 deficiency	611126	●
BCS1L	2q35	Mitochondrial complex III deficiency, nuclear type 1	124000	●
UQCRB	8q22.1	Mitochondrial complex III deficiency, nuclear type 3	615158	●
UQCRQ	5q31.1	Mitochondrial complex III deficiency, nuclear type 4	615159	●
COX10	17p12	Mitochondrial complex IV deficiency	220110	●
COX6B1	19q13.12	Mitochondrial complex IV deficiency	220110	●
FASTKD2	2q33.3	Mitochondrial complex IV deficiency	220110	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
TYMP	22q13.33	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	●
TK2	16q21	Mitochondrial DNA depletion syndrome 2 (myopathic type)	609560	●
DGUOK	2p13.1	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	251880	●
POLG	15q26.1	Mitochondrial DNA depletion syndrome 4A (Alpers type)	203700	●
POLG	15q26.1	Mitochondrial DNA depletion syndrome 4B (MNGIE type)	613662	●
SUCLA2	13q14.2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	●
MPV17	2p23.3	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	256810	●
C10orf2	10q24.31	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	271245	●
RRM2B	8q22.3	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075	●
RRM2B	8q22.3	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075	●
SUCLG1	2p11.2	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	245400	●
POLG	15q26.1	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	607459	●
TIMM8A	Xq22.1	Mohr-Tranebjaerg syndrome	304700	●
MOCS1	6p21.2	Molybdenum cofactor deficiency A	252150	●
MOCS2	5q11.2	Molybdenum cofactor deficiency B	252160	●
ZEB2	2q22.3	Mowat-Wilson syndrome	235730	●
GNPTAB	12q23.2	Mucolipidosis II alpha/beta	252500	●
GNPTAB	12q23.2	Mucolipidosis III alpha/beta	252600	●
MCOLN1	19p13.2	Mucolipidosis IV	252650	●
SGSH	17q25.3	Mucopolysaccharidisis type IIIA (Sanfilippo A)	252900	●
IDUA	4p16.3	Mucopolysaccharidosis Ih (Hurler Syndrome)	607014	●
IDUA	4p16.3	Mucopolysaccharidosis Ih/s (HURLER-SCHEIE SYNDROME)	607015	●
IDS	Xq28	Mucopolysaccharidosis II	309900	●
IDUA	4p16.3	Mucopolysaccharidosis Is (SCHEIE SYNDROME)	607016	●
NAGLU	17q21.2	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	●
HGSNAT	8p11.21	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930	●
GLB1	3p22.3	Mucopolysaccharidosis type IVB (Morquio)	253010	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
ARSB	5q14.1	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200	●
GUSB	7q11.21	Mucopolysaccharidosis VII	253220	●
TRIM37	17q22	Mulibrey nanism	253250	●
CHRNA1	2q31.1	Multiple pterygium syndrome, lethal type	253290	●
CHRND	2q37.1	Multiple pterygium syndrome, lethal type	253290	●
CHRNG	2q37.1	Multiple pterygium syndrome, lethal type	253290	●
PLEC	8q24.3	Muscular dystrophy with epidermolysis bullosa simplex	226670	●
LMNA	1q22	Muscular dystrophy, congenital	613205	●
LAMA2	6q22.33	Muscular dystrophy, congenital merosin-deficient	607855	●
LAMA2	6q22.33	Muscular dystrophy, congenital, due to partial LAMA2 deficiency	607855	●
LMNA	1q22	Muscular dystrophy, limb-girdle, type 1B	159001	●
PLEC	8q24.3	Muscular dystrophy, limb-girdle, type 2Q	613723	●
SEPN1	1p36.11	Muscular dystrophy, rigid spine, 1	602771	●
POMT1	9q34.13	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	236670	●
POMT2	14q24.3	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	613150	●
POMGNT1	1p34.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	253280	●
FKTN	9q31.2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	253800	●
FKRP	19q13.32	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153	●
LARGE	22q12.3	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	613154	●
POMT1	9q34.13	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	613155	●
POMT2	14q24.3	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	613156	●
POMGNT1	1p34.1	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	613151	●
LARGE	22q12.3	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	608840	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
FKRP	19q13.32	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	606612	●
FKTN	9q31.2	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	613152	●
POMT1	9q34.13	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	609308	●
POMT2	14q24.3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	613158	●
POMGNT1	1p34.1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	613157	●
FKTN	9q31.2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	611588	●
FKRP	19q13.32	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	607155	●
DOK7	4p16.3	Myasthenic syndrome, congenital, 10	254300	●
RAPSN	11p11.2	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	616326	●
DPAGT1	11q23.3	Myasthenic syndrome, congenital, 13, with tubular aggregates	614750	●
ALG2	9q22.33	Myasthenic syndrome, congenital, 14, with tubular aggregates	616228	●
CHRNA1	2q31.1	Myasthenic syndrome, congenital, 1A, slow-channel	601462	●
CHRNA1	2q31.1	Myasthenic syndrome, congenital, 1B, fast-channel	608930	●
CHRND	2q37.1	Myasthenic syndrome, congenital, 3A, slow-channel	616321	●
CHRND	2q37.1	Myasthenic syndrome, congenital, 3B, fast-channel		●
CHRND	2q37.1	Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	616323	●
MPL	1p34.2	Myelofibrosis with myeloid metaplasia, somatic	254450	●
CPT2	1p32.3	Myopathy due to CPT II deficiency	255110	●
SEPN1	1p36.11	Myopathy, congenital, with fiber-type disproportion	255310	●
STIM1	11p15.4	Myopathy, tubular aggregate, 1	160565	●
ORAI1	12q24.31	Myopathy, tubular aggregate, 2	615883	●
SCO2	22q13.33	Myopia 6	608908	●
COL6A2	21q22.3	Myosclerosis, congenital	255600	●
MTM1	Xq28	Myotubular myopathy, X-linked	310400	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
NAGS	17q21.31	N-acetylglutamate synthase deficiency	237310	●
NHS	Xp22.13	Nance-Horan syndrome	302350	●
NEB	2q23.3	Nemaline myopathy 2, autosomal recessive	256030	●
CLCN5	Xp11.23-p11.22	Nephrolithiasis, type I	310468	●
NPHP1	2q13	Nephronophthisis 1, juvenile	256100	●
TMEM67	8q22.1	Nephronophthisis 11	613550	●
INVS	9q31.1	Nephronophthisis 2, infantile	602088	●
NPHP3	3q22.1	Nephronophthisis 3	604387	●
NPHP4	1p36.31	Nephronophthisis 4	606966	●
NPHS1	19q13.12	Nephrotic syndrome, type 1	256300	●
NPHS2	1q25.2	Nephrotic syndrome, type 2	600995	●
PLCE1	10q23.33	Nephrotic syndrome, type 3	610725	●
LAMB2	3p21.31	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199	●
FOLR1	11q13.4	Neurodegeneration due to cerebral folate transport deficiency	613068	●
PLA2G6	22q13.1	Neurodegeneration with brain iron accumulation 2B	610217	●
IGHMBP2	11q13.3	Neuronopathy, distal hereditary motor, type VI	604320	●
MPZ	1q23.3	Neuropathy, congenital hypomyelinating	605253	●
EGR2	10q21.3	Neuropathy, congenital hypomyelinating, 1	605253	●
PMP22	17p12	Neuropathy, inflammatory demyelinating	139393	●
PMP22	17p12	Neuropathy, recurrent, with pressure palsies	162500	●
HAX1	1q21.3	Neutropenia, severe congenital 3, autosomal recessive	610738	●
G6PC3	17q21.31	Neutropenia, severe congenital 4, autosomal recessive	612541	●
WAS	Xp11.23	Neutropenia, severe congenital, X-linked	300299	●
SMPD1	11p15.4	Niemann-Pick disease, type A	257200	●
SMPD1	11p15.4	Niemann-Pick disease, type B	607616	●
NPC1	18q11.2	Niemann-Pick disease, type C1	257220	●
NPC2	14q24.3	Niemann-pick disease, type C2	607625	●
NPC1	18q11.2	Niemann-Pick disease, type D	257220	●
NBN	8q21.3	Nijmegen breakage syndrome	251260	●
CASK	Xp11.4	Non-Hodgkin lymphoma, somatic	605027	●
NDP	Xp11.3	Norrie disease	310600	●
NXF5	Xq22.1	Nuclear RNA export factor 5	300319	●
ENPP1	6q23.2	Obesity, susceptibility to	601665	●
ATP7A	Xq21.1	Occipital horn syndrome	304150	●
ALPL	1p36.12	Odontohypophosphatasia	146300	●
WNT10A	2q35	Odontoonychodermal dysplasia	257980	●
MED12	Xq13.1	Ohdo syndrome, X-linked	300895	●
MBTPS2	Xp22.12-p22.11	Olmsted syndrome, X-linked	300918	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
DCLRE1C	10p13	Omenn syndrome	603554	●
RAG1	11p12	Omenn syndrome	603554	●
RAG2	11p12	Omenn syndrome	603554	●
MID1	Xp22.2	Opitz GBBB syndrome, type I	300000	●
MED12	Xq13.1	Opitz-Kaveggia syndrome	305450	●
OPA3	19q13.32	Optic atrophy 3 with cataract	165300	●
OTC	Xp11.4	Ornithine transcarbamylase deficiency	311250	●
OFD1	Xp22.2	Orofaciodigital syndrome I	311200	●
COL1A1	17q21.33	Osteogenesis imperfecta, type I	166200	●
COL1A1	17q21.33	Osteogenesis imperfecta, type II	166210	●
COL1A2	7q21.3	Osteogenesis imperfecta, type II	166210	●
COL1A1	17q21.33	Osteogenesis imperfecta, type III	259420	●
COL1A2	7q21.3	Osteogenesis imperfecta, type III	259420	●
COL1A1	17q21.33	Osteogenesis imperfecta, type IV	166220	●
COL1A2	7q21.3	Osteogenesis imperfecta, type IV	166220	●
CRTAP	3p22.3	Osteogenesis imperfecta, type VII	610682	●
LEPRE1	1p34.2	Osteogenesis imperfecta, type VIII	610915	●
CLCN7	16p13.3	Osteopetrosis, autosomal dominant 2	166600	●
TCIRG1	11q13.2	Osteopetrosis, autosomal recessive 1	259700	●
CA2	8q21.2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	259730	●
CLCN7	16p13.3	Osteopetrosis, autosomal recessive 4	611490	●
OSTM1	6q21	Osteopetrosis, autosomal recessive 5	259720	●
VDR	12q13.11	Osteoporosis, involutional	166710	●
COL1A2	7q21.3	Osteoporosis, postmenopausal	166710	●
TNFRSF11B	8q24.12	Paget disease of bone 5, juvenile-onset	239000	●
SOX3	Xq27.1	Panhypopituitarism, X-linked	312000	●
PLA2G6	22q13.1	Parkinson disease 14, autosomal recessive	612953	●
ARX	Xp21.3	Partington syndrome	309510	●
LBR	1q42.12	Pelger-Huet anomaly	169400	●
PLP1	Xq22.2	Pelizaeus-Merzbacher disease	312080	●
ACOX1	17q25.1	Peroxisomal acyl-CoA oxidase deficiency	264470	●
PEX13	2p16.1	Peroxisome biogenesis disorder 11 B	614885	●
PEX13	2p16.1	Peroxisome biogenesis disorder 11A (Zellweger	614883	●
PEX1	7q21.2	Peroxisome biogenesis disorder 1A (Zellweger)	214100	●
PEX1	7q21.2	Peroxisome biogenesis disorder 1B (NALD/IRD)	601539	●
PEX12	17q12	Peroxisome biogenesis disorder 3A (Zellweger	614859	●
PEX12	17q12	Peroxisome biogenesis disorder 3B	266510	●
PEX10	1p36.32	Peroxisome biogenesis disorder 6A (Zellweger)	614870	●
PEX10	1p36.32	Peroxisome biogenesis disorder 6B	614871	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
PEX26	22q11.21	Peroxisome biogenesis disorder 7A (Zellweger)	614872	●
PEX26	22q11.21	Peroxisome biogenesis disorder 7B	614873	●
PEX5	3q26.33	Peroxisome biogenesis disorder 7B	611058	●
PEX7	6q23.3	Peroxisome biogenesis disorder 9B	614879	●
HSD17B4	5q23.1	Perrault syndrome 1	233400	●
C10orf2	10q24.31	Perrault syndrome 5		●
PAH	12q23.2	Phenylketonuria	261600	●
PRPS1	Xq22.3	Phosphoribosylpyrophosphate synthetase superactivity	300661	●
LAMB2	3p21.31	Pierson syndrome	609049	●
TCF4	18q21.2	Pitt-Hopkins syndrome	610954	●
POU1F1	3p11.2	Pituitary hormone deficiency, combined, 1	613038	●
PROP1	5q35.3	Pituitary hormone deficiency, combined, 2	262600	●
LHX3	9q34.3	Pituitary hormone deficiency, combined, 3	221750	●
HESX1	3p14.3	Pituitary hormone deficiency, combined, 5	182230	●
PLG	6q26	Plasminogen deficiency, type I	217090	●
PKHD1	6p12.3-p12.2	Polycystic kidney and hepatic disease	263200	●
GBE1	3p12.2	Polyglucosan body disease, adult form	263570	●
TSEN54	17q25.1	Pontocerebellar hypoplasia type 2A	277470	●
TSEN54	17q25.1	Pontocerebellar hypoplasia type 4	225753	●
TSEN54	17q25.1	Pontocerebellar hypoplasia type 5	610204	●
MVK	12q24.11	Porokeratosis 3, disseminated superficial actinic	175900	●
UROS	10q26.1-q26.2	Porphyria, congenital erythropoietic	263700	●
LHCGR	2p16.3	Precocious puberty, male	176410	●
NR5A1	9q33.3	Premature ovarian failure 7	612964	●
ALS2	2q33.1	Primary lateral sclerosis, juvenile	606353	●
POLG	15q26.1	Progressive external ophthalmoplegia, autosomal recessive	258450	●
CFP	Xp11.23	Properdin deficiency, X-linked	312060	●
PCCA	13q32.3	Propionicacidemia	606054	●
PCCB	3q22.3	Propionicacidemia	606054	●
CLCN5	Xp11.23-p11.22	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	308990	●
ARX	Xp21.3	Proud syndrome	300004	●
HSD17B3	9q22.32	Pseudohermaphroditism, male, with gynecomastia	264300	●
SCNN1A	12p13.31	Pseudohypoaldosteronism, type I	264350	●
SCNN1B	16p12.2	Pseudohypoaldosteronism, type I	264350	●
SCNN1G	16p12.2	Pseudohypoaldosteronism, type I	264350	●
SRD5A2	2p23.1	Pseudovaginal perineoscrotal hypospadias	264600	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
CTSK	1q21.3	Pycnodysostosis	265800	🔴
MYD88	3p22.2	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260	🔴
PNPO	17q21.32	Pyridoxamine 5'-phosphate oxidase deficiency	610090	🔴
PC	11q13.2	Pyruvate carboxylase deficiency	266150	🔴
PDHA1	Xp22.12	Pyruvate dehydrogenase E1-alpha deficiency	312170	🔴
PDP1	8q22.1	Pyruvate dehydrogenase phosphatase deficiency	608782	🔴
PKLR	1q22	Pyruvate kinase deficiency	266200	🔴
INSR	19p13.2	Rabson-Mendenhall syndrome	262190	🔴
FAM20C	7p22.3	Raine syndrome	259775	🔴
NPHP3	3q22.1	Renal-hepatic-pancreatic dysplasia 1	208540	🔴
PQBP1	Xp11.23	Renpenning syndrome	309500	🔴
ZMPSTE24	1p34.2	Restrictive dermopathy, lethal	275210	🔴
LMNA	1q22	Restrictive dermopathy, lethal	275210	🔴
OFD1	Xp22.2	Retinitis pigmentosa 23	300424	🔴
USH2A	1q41	Retinitis pigmentosa 39	613809	🔴
CLRN1	3q25.1	Retinitis pigmentosa 61	614180	🔴
MECP2	Xq28	Rett syndrome	312750	🔴
FOXG1	14q12	Rett syndrome, congenital variant	613454	🔴
MECP2	Xq28	Rett syndrome, preserved speech variant	312750	🔴
LBR	1q42.12	Reynolds syndrome	613471	🔴
PEX7	6q23.3	Rhizomelic chondrodysplasia punctata, type 1	215100	🔴
AGPS	2q31.2	Rhizomelic chondrodysplasia punctata, type 3	600121	🔴
VDR	12q13.11	Rickets, vitamin D-resistant, type IIA	277440	🟡
ESCO2	8p21.1	Roberts syndrome	268300	🔴
MPZ	1q23.3	Roussy-Levy syndrome	180800	🔴
PMP22	17p12	Roussy-Levy syndrome	180800	🔴
SLC17A5	6q13	Salla disease	604369	🔴
HEXB	5q13.3	Sandhoff disease, infantile, juvenile, and adult forms	268800	🔴
ESCO2	8p21.1	SC phocomelia syndrome	269000	🔴
SLC35D1	1p31.3	Schneckenbecken dysplasia	269250	🔴
WNT10A	2q35	Schopf-Schulz-Passarge syndrome	224750	🟡
HSPG2	1p36.12	Schwartz-Jampel syndrome, type 1	255800	🔴
JAK3	19p13.11	SCID, autosomal recessive, T-negative/B-positive type	600802	🔴
ATR	3q23	Seckel syndrome 1	210600	🔴
TH	11p15.5	Segawa syndrome, recessive	605407	🔴
NPHP4	1p36.31	Senior-Loken syndrome 4	606996	🔴
IQCB1	3q13.33	Senior-Loken syndrome 5	609254	🔴
CEP290	12q21.32	Senior-Loken syndrome 6	610189	🔴
NPHP1	2q13	Senior-Loken syndrome-1	266900	🔴
HESX1	3p14.3	Septooptic dysplasia	182230	🔴

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
ADA	20q13.12	Severe combined immunodeficiency due to ADA deficiency	102700	●
NHEJ1	2q35	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	●
DCLRE1C	10p13	Severe combined immunodeficiency, Athabascan type	602450	●
RAG1	11p12	Severe combined immunodeficiency, B cell-negative	601457	●
RAG2	11p12	Severe combined immunodeficiency, B cell-negative	601457	●
IL2R	Xq13.1	Severe combined immunodeficiency, X-linked	300400	●
IFT80	3q25.33	Short-rib thoracic dysplasia 2 with or without polydactyly	611263	●
DYNC2H1	11q22.3	Short-rib thoracic dysplasia 3 with or without polydactyly	613091	●
SBDS	7q11.21	Shwachman-Bodian-Diamond syndrome	260400	●
SLC17A5	6q13	Sialic acid storage disorder, infantile	269920	●
NEU1	6p21.33	Sialidosis, type I	256550	●
NEU1	6p21.33	Sialidosis, type II	256550	●
HBB	11p15.4	Sickle cell anemia	603903	●
GPC3	Xq26.2	Simpson-Golabi-Behmel syndrome, type 1	312870	●
OFD1	Xp22.2	Simpson-Golabi-Behmel syndrome, type 2	300209	●
ALDH3A2	17p11.2	Sjogren-Larsson syndrome	270200	●
DSP	6p24.3	Skin fragility-woolly hair syndrome	607655	●
DHCR7	11q13.4	Smith-Lemli-Opitz syndrome	270400	●
NSD1	5q35.2-q35.3	Sotos syndrome 1	117550	●
SACS	13q12.12	Spastic ataxia, Charlevoix-Saguenay type	270550	●
ALS2	2q33.1	Spastic paralysis, infantile onset ascending	607225	●
PLP1	Xq22.2	Spastic paraplegia 2, X-linked	312920	●
GJC2	1q42.13	Spastic paraplegia 44, autosomal recessive	613206	●
NR5A1	9q33.3	Spermatogenic failure 8	613957	●
AR	Xq12	Spinal and bulbar muscular atrophy of Kennedy	313200	●
PLEKHG5	1p36.31	Spinal muscular atrophy, distal, autosomal recessive, 4	611067	●
ATP7A	Xq21.1	Spinal muscular atrophy, distal, X-linked 3	300489	●
UBA1	Xp11.23	Spinal muscular atrophy, X-linked 2, infantile	301830	●
SMN1	5q13.2	Spinal muscular atrophy-1	253300	●
SMN1	5q13.2	Spinal muscular atrophy-2	253550	●
SMN1	5q13.2	Spinal muscular atrophy-3	253400	●
SMN1	5q13.2	Spinal muscular atrophy-4	271150	●
TPP1	11p15.4	Spinocerebellar ataxia, autosomal recessive 7	609270	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
DLL3	19q13.2	Spondylocostal dysostosis 1, autosomal recessive	277300	●
FAS	10q23.31	Squamous cell carcinoma, burn scar-related, somatic		●
SHROOM4	Xp11.22	Stocco dos Santos X-linked mental retardation syndrome	300434	●
STIM1	11p15.4	Stormorken syndrome	185070	●
NUP62	19q13.33	Striatonigral degeneration, infantile	271930	●
LIFR	5p13.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559	●
DCX	Xq23	Subcortical laminal heteropia, X-linked	300067	●
ALDH5A1	6p22.3	Succinic semialdehyde dehydrogenase deficiency	271980	●
OXCT1	5p13.1	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	●
TSPYL1	6q22.1	Sudden infant death with dysgenesis of the testes syndrome	608800	●
SUOX	12q13.2	Sulfite oxidase deficiency	272300	●
SFTPB	2p11.2	Surfactant metabolism dysfunction, pulmonary, 1	265120	●
SFTPC	8p21.3	Surfactant metabolism dysfunction, pulmonary, 2	610913	●
ABCA3	16p13.3	Surfactant metabolism dysfunction, pulmonary, 3	610921	●
TREX1	3p21.31	Systemic lupus erythematosus, susceptibility to	152700	●
HEXA	15q23	Tay-Sachs disease	272800	●
FOXN1	17q11.2	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	601705	●
WNT3	17q21.31	Tetra-amelia syndrome	273395	●
HBA1	16p13.3	Thalassemias, alpha-	604131	●
HBB	11p15.4	Thalassemias, beta-	613985	●
MPL	1p34.2	Thrombocythemia 2	601977	●
MPL	1p34.2	Thrombocytopenia, congenital amegakaryocytic	604498	●
WAS	Xp11.23	Thrombocytopenia, X-linked	313900	●
WAS	Xp11.23	Thrombocytopenia, X-linked, intermittent	313900	●
F9	Xq27.1	Thrombophilia, X-linked, due to factor IX defect	300807	●
CBS	21q22.3	Thrombosis, hyperhomocysteinemic	236200	●
ADAMTS13	9q34.2	Thrombotic thrombocytopenic purpura, familial	274150	●
COL7A1	3p21.31	Toenail dystrophy, isolated	607523	●
WNT10A	2q35	Tooth agenesis, selective, 4	150400	●
EDA	Xq13.1	Tooth agenesis, selective, X-linked 1	313500	●
COL7A1	3p21.31	Transient bullous of the newborn	131705	●

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM	
ERCC2	19q13.32	Trichothiodystrophy 1, photosensitive	601675	●
ERCC3	2q14.3	Trichothiodystrophy 2, photosensitive	616390	●
GTF2H5	6q25.3	Trichothiodystrophy 3, photosensitive	616395	●
HADHA	2p23.3	Trifunctional protein deficiency	609015	●
HADHB	2p23.3	Trifunctional protein deficiency	609015	●
FAH	15q25.1	Tyrosinemia, type I	276700	●
TAT	16q22.2	Tyrosinemia, type II	276600	●
HP	12q24.31	Tyrosinemia, type III	276710	●
COL6A1	21q22.3	Ullrich congenital muscular dystrophy	254090	●
COL6A2	21q22.3	Ullrich congenital muscular dystrophy	254090	●
COL6A3	2q37.3	Ullrich congenital muscular dystrophy	254090	●
WNT7A	3p25.1	Ulna and fibula, absence of, with severe limb deficiency	276820	●
MYO7A	11q13.5	Usher syndrome, type 1B	276900	●
USH1C	11p15.1	Usher syndrome, type 1C	276904	●
CDH23	10q22.1	Usher syndrome, type 1D	601067	●
CDH23	10q22.1	Usher syndrome, type 1D/F digenic	601067	●
USH1G	17q25.1	Usher syndrome, type 1G	606943	●
USH2A	1q41	Usher syndrome, type 2A	276901	●
GPR98	5q14.3	Usher syndrome, type 2C	605472	●
GPR98	5q14.3	Usher syndrome, type 2C, GPR98/PDZD7 digenic	605472	●
CLRN1	3q25.1	Usher syndrome, type 3A	276902	●
ERCC6	10q11.23	UV-sensitive syndrome 1	600630	●
ERCC8	5q12.1	UV-sensitive syndrome 2	614621	●
ZIC3	Xq26.3	VACTERL association, X-linked	314390	●
TREX1	3p21.31	Vasculopathy, retinal, with cerebral leukodystrophy	192315	●
PTEN	10q23.31	VATER association with macrocephaly and ventriculomegaly	276950	●
CYP27B1	12q14.1	Vitamin D-dependent rickets, type I	264700	●
ACADL	17p13.1	VLCAD deficiency	201475	●
ACADVL	17p13.1	VLCAD deficiency	201475	●
GJB2	13q12.11	Vohwinkel syndrome	124500	●
EDNRB	13q22.3	Waardenburg syndrome, type 4A	277580	●
EDN3	20q13.32	Waardenburg syndrome, type 4B	613265	●
RAB39B	Xq28	Waisman syndrome	311510	●
RAB3GAP1	2q21.3	Warburg micro syndrome 1	600118	●
RAB3GAP2	1q41	Warburg micro syndrome 2	614225	●
F9	Xq27.1	Warfarin sensitivity	122700	●
EVC	4p16.2	Weyers acrodental dysostosis	193530	●
EVC2	4p16.2	Weyers acrofacial dysostosis	193530	●
GPC3	Xq26.2	Wilms tumor, somatic	194070	●
ATP7B	13q14.3	Wilson disease	277900	●
WAS	Xp11.23	Wiskott-Aldrich syndrome	301000	●

COMPATIBILITY

GENETIC TEST

GENE	LOCUS	DISEASE	OMIM	
EIF2AK3	2p11.2	Wolcott-Rallison syndrome	226980	🔴
ATP6V0A2	12q24.31	Wrinkly skin syndrome	278250	🟡
XPA	9q22.33	Xeroderma pigmentosum, group A	278700	🔴
ERCC3	2q14.3	Xeroderma pigmentosum, group B	610651	🔴
XPC	3p25.1	Xeroderma pigmentosum, group C	278720	🔴
ERCC2	19q13.32	Xeroderma pigmentosum, group D	278730	🔴
DDB2	11p11.2	Xeroderma pigmentosum, group E, DDB-negative subtype	278740	🔴
ERCC4	16p13.12	Xeroderma pigmentosum, group F	278760	🔴
ERCC5	3q33.1	Xeroderma pigmentosum, group G	278780	🔴
ERCC5	3q33.1	Xeroderma pigmentosum, group G/Cockayne syndrome	278780	🔴
ERCC4	16p13.12	Xeroderma pigmentosum, type F/Cockayne syndrome	278760	🔴
ERCC4	16p13.12	XFE progeroid syndrome	610965	🔴
ZNF41	Xp11.23	Zinc Finger Protein 41	*314995*	🔴
ZNF674	Xp11.3-p11.2	Zinc Finger Protein 674	*300573*	🔴