COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

13 NEW RECESSIVE DISEASES
detected by the Genetic Compatibility Test (GCT), from 1st of July 2020

SERPINA1	14q32.13	Alpha-1-antitrypsin deficiency	613490
SPG7	16q24.3	Autosomal recessive spastic paraplegia type 7	607259
OCA2	15q12	Oculocutaneous albinism type 2	203200
SLC26A4	7q22.3	Autosomal recessive non-syndromic sensorineural deafness type DFNB/Pendred syndrome	274600
CLCN1	7q34	Thomsen and Becker disease	160800
TYR	11q14.3	Oculocutaneous albinism type 1A	203100
CAPN3	15q15.1	Calpain-3-related limb-girdle muscular dystrophy R1	253600
CRB1	17p13.1	Leber congenital amaurosis	204000
GALNS	16q24.3	Mucopolysaccharidosis type 4A	253000
OTOF	2p23.3	Autosomal recessive non-syndromic sensorineural deafness type DFNB	601071
SAG	2q37.1	Retinitis pigmentosa 47	613758
TMPRSS3	21q22.3	Deafness, autosomal recessive 8/10	601072
TSHR	14q31.1	Hyperthyroidism	603373

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

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GENE	LOCUS	DISEASE	OMIM
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
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

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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
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

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GENE	LOCUS	DISEASE	OMIM
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
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

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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
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

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GENE	LOCUS	DISEASE	OMIM
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
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

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GENE	LOCUS	DISEASE	OMIM
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
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

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GENE	LOCUS	DISEASE	OMIM
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
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

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GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	LOCUS	DISEASE	OMIM
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
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*