Tuset, 23 6º1ª
08006-Barcelona
heres@fullgenomics.es

#	Diseases	GENES	INHERITANCE
99	Ehlers-Danlos Syndrome: Type VIIC	ADAMTS2	AR
100	Ellis-van Creveld Syndrome: EVC Related	EVC	AR
101	Ellis-van Creveld Syndrome: EVC2 Related	EVC,EVC2	AR
102	Emery-Dreifuss Myopathy: X-Linked	EMD	XL
103	Enhanced S-Cone	NR2E3	AR
104	Ethylmalonic Aciduria	ETHE1	AR
105	Fabry's Disease	GLA	XL
106	Factor IX Deficiency	F9	XL
107	Factor VIII Deficiency	F8	XL
108	Familial Chloride Diarrhea	SLC26A3	AR
109	Familial Dysautonomia	IKBKAP	AR
110	Familial Hyperinsulinism: Type 1: ABCC8 Related	ABCC8	AR
111	Familial Hyperinsulinism: Type 2: KCNJ11 Related	KCNJ11	AR
112	Familial Mediterranean Fever	MEFV	AR
113	Fanconi Anemia: Type A	FANCA	AR
114	Fanconi Anemia: Type C	FANCC	AR
115	Fanconi Anemia: Type G	FANCG	AR
116	Fanconi Anemia: Type J	BRIP1	AR
117	Fragile X Syndrome	FMR1	XL
118	Fumarase Deficiency	FH	AR
119	GM1-Gangliosidoses	GLB1	AR
120	GRACILE Syndrome	BCS1L	AR
121	Galactokinase Deficiency	GALK1	AR
122	Gaucher Disease	GBA	AR
123	Gitelman Syndrome	SLC12A3	AR
124	Globoid Cell Leukodystrophy	GALC	AR
125	Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	XL
126	Glutaric Acidemia: Type I	GCDH	AR
127	Glutaric Acidemia: Type IIA	ETFA	AR
128	Glutaric Acidemia: Type IIB	ETFB	AR
129	Glutaric Acidemia: Type IIC	ETFDH	AR
130	Glycine Encephalopathy: AMT Related	AMT	AR
131	Glycine Encephalopathy: GLDC Related	GLDC	AR
132	Glycogen Storage Disease: Type IA	G6PC	AR
133	Glycogen Storage Disease: Type IB	SLC37A4	AR
134	Glycogen Storage Disease: Type II	GAA	AR
135	Glycogen Storage Disease: Type III	AGL	AR
136	Glycogen Storage Disease: Type IV	GBE1	AR
137	Glycogen Storage Disease: Type V	PYGM	AR
138	Glycogen Storage Disease: Type VII	PFKM	AR
139	Guanidinoacetate Methyltransferase Deficiency	GAMT	AR
140	HMG-CoA Lyase Deficiency	HMGCL	AR
141	Hemochromatosis: Type 2A: HFE2 Related	HFE2	AR
142	Hemochromatosis: Type 3: TFR2 Related	TFR2	AR
143	Hemoglobinopathy: Hb C	HBB	AR
144	Hemoglobinopathy: Hb D	HBB	AR
145	Hemoglobinopathy: Hb E	HBB	AR
146	Hemoglobinopathy: Hb O	HBB	AR
147	Hereditary Fructose Intolerance	ALDOB	AR

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148	Hereditary Spastic Paraplegia: TECPR2 Related	TECPR2	AR
149	Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related	LAMA3	AR
150	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	LAMB3	AR
151	Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related	LAMC2	AR
152	Hermansky-Pudlak Syndrome: Type 1	HPS1	AR
153	Hermansky-Pudlak Syndrome: Type 3	HPS3	AR
154	Hermansky-Pudlak Syndrome: Type 4	HPS4	AR
155	Holocarboxylase Synthetase Deficiency	HLCS	AR
156	Homocystinuria Caused by CBS Deficiency	CBS	AR
157	Hunter Syndrome	IDS	XL
158	Hurler Syndrome	IDUA	AR
159	Hypohidrotic Ectodermal Dysplasia: X-Linked	EDA	XL
160	Hypophosphatasia	ALPL	AR
161	Inclusion Body Myopathy: Type 2	GNE	AR
162	Infantile Cerebral and Cerebellar Atrophy	MED17	AR
163	Isolated Microphthalmia: VSX2 Related	VSX2	AR
164	Isovaleric Acidemia	IVD	AR
165	Joubert Syndrome	TMEM216	AR
166	Juvenile Retinoschisis: X-Linked	RS1	XL
167	Lamellar Ichthyosis: Type 1	TGM1	AR
168	Laryngoonychocutaneous Syndrome	LAMA3	AR
169	Leber Congenital Amaurosis: CEP290 Related	CEP290	AR
170	Leber Congenital Amaurosis: GUCY2D Related	GUCY2D	AR
171	Leber Congenital Amaurosis: LCA5 Related	LCA5	AR
172	Leber Congenital Amaurosis: RDH12 Related	RDH12	AR
173	Leigh Syndrome: French-Canadian	LRPPRC	AR
174	Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related	EIF2B5	AR
175	Leydig Cell Hypoplasia (Luteinizing Hormone Resistance)	LHCGR	AR
176	Limb-Girdle Muscular Dystrophy: Type 2A	CAPN3	AR
177	Limb-Girdle Muscular Dystrophy: Type 2B	DYSF	AR
178	Limb-Girdle Muscular Dystrophy: Type 2C	SGCG	AR
179	Limb-Girdle Muscular Dystrophy: Type 2D	SGCA	AR
180	Limb-Girdle Muscular Dystrophy: Type 2E	SGCB	AR
181	Limb-Girdle Muscular Dystrophy: Type 2F	SGCD	AR
182	Limb-Girdle Muscular Dystrophy: Type 2I	FKRP	AR
183	Lipoprotein Lipase Deficiency	LPL	AR
184	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	AR
185	Lowe Oculocerebrorenal Syndrome	OCRL	XL
186	Lysinuric Protein Intolerance	SLC7A7	AR
187	Malonyl-CoA Decarboxylase Deficiency	MLYCD	AR
188	Maple Syrup Urine Disease: Type 1A	BCKDHA	AR
189	Maple Syrup Urine Disease: Type 1B	BCKDHB	AR
190	Maple Syrup Urine Disease: Type 2	DBT	AR
191	Maple Syrup Urine Disease: Type 3	DLD	AR
192	Maroteaux-Lamy Syndrome	ARSB	AR
193	Meckel Syndrome: Type 1	MKS1	AR
194	Medium-Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	AR
195	Megalencephalic Leukoencephalopathy	MLC1	AR
196	Metachromatic Leukodystrophy	ARSA	AR

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197	Methylmalonic Acidemia: MMAA Related	MMAA	AR
198	Methylmalonic Acidemia: MMAB Related	MMAB	AR
199	Methylmalonic Acidemia: MUT Related	MUT	AR
200	Methylmalonic Aciduria and Homocystinuria: Type cblC	MMACHC	AR
201	Mitochondrial Complex I Deficiency: NDUFS6 Related	NDUFS6	AR
202	Mitochondrial DNA Depletion Syndrome: MNGIE Type	TYMP	AR
203	Mitochondrial Myopathy and Sideroblastic Anemia	PUS1	AR
204	Mitochondrial Trifunctional Protein Deficiency: HADHB Related	HADHB	AR
205	Morquio Syndrome: Type A	GALNS	AR
206	Morquio Syndrome: Type B	GLB1	AR
207	Mucolipidosis: Type II/III	GNPTAB	AR
208	Mucolipidosis: Type IV	MCOLN1	AR
209	Multiple Pterygium Syndrome	CHRNG	AR
210	Multiple Sulfatase Deficiency	SUMF1	AR
211	Muscle-Eye-Brain Disease	POMGNT1	AR
212	Myotubular Myopathy: X-Linked	MTM1	XL
213	Navajo Neurohepatopathy	MPV17	AR
214	Nemaline Myopathy: NEB Related	NEB	AR
215	Nephrotic Syndrome: Type 1	NPHS1	AR
216	Nephrotic Syndrome: Type 2	NPHS2	AR
217	Neuronal Ceroid-Lipofuscinosis: CLN5 Related	CLN5	AR
218	Neuronal Ceroid-Lipofuscinosis: CLN6 Related	CLN6	AR
219	Neuronal Ceroid-Lipofuscinosis: CLN8 Related	CLN8	AR
220	Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	MFSD8	AR
221	Neuronal Ceroid-Lipofuscinosis: PPT1 Related	PPT1	AR
222	Neuronal Ceroid-Lipofuscinosis: TPP1 Related	TPP1	AR
223	Niemann-Pick Disease: Type A	SMPD1	AR
224	Niemann-Pick Disease: Type B	SMPD1	AR
225	Niemann-Pick Disease: Type C1	NPC1	AR
226	Niemann-Pick Disease: Type C2	NPC2	AR
227	Nijmegen Breakage Syndrome	NBN	AR
228	Nonsyndromic Hearing Loss and Deafness: GJB2 Related	GJB2	AR
229	Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	LOXHD1	AR
230	Nonsyndromic Hearing Loss and Deafness: MYO15A Related	MYO15A	AR
231	Oculocutaneous Albinism: Type 1	TYR	AR
232	Oculocutaneous Albinism: Type 3	TYRP1	AR
233	Oculocutaneous Albinism: Type 4	SLC45A2	AR
234	Omenn Syndrome: DCLRE1C Related	DCLRE1C	AR
235	Omenn Syndrome: RAG2 Related	RAG2	AR
236	Ornithine Transcarbamylase Deficiency	OTC	AR
237	Ornithine Translocase Deficiency	SLC25A15	XL
238	Osteopetrosis: TCIRG1 Related	TCIRG1	AR
239	POLG Related Disorders: Autosomal Recessive	POLG	AR
240	Papillon-Lefevre Syndrome	CTSC	AR
241	Pendred Syndrome	SLC26A4	AR
242	Persistent Mullerian Duct Syndrome: Type I	AMH	AR
243	Persistent Mullerian Duct Syndrome: Type II	AMHR2	AR
244	Phenylalanine Hydroxylase Deficiency	PAH	AR
245	Polyglandular Autoimmune Syndrome: Type I	AIRE	AR

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246	Pontocerebellar Hypoplasia: EXOSC3 Related	EXOSC3	AR
247	Pontocerebellar Hypoplasia: RARS2 Related	RARS2	AR
248	Pontocerebellar Hypoplasia: SEPSECS Related	SEPSECS	AR
249	Pontocerebellar Hypoplasia: TSEN54 Related	TSEN54	AR
250	Pontocerebellar Hypoplasia: VPS53 Related	VPS53	AR
251	Pontocerebellar Hypoplasia: VRK1 Related	VRK1	AR
252	Primary Carnitine Deficiency	SLC22A5	AR
253	Primary Ciliary Dyskinesia: DNAI1 Related	DNAI1	AR
254	Primary Ciliary Dyskinesia: DNAI2 Related	DNAI2	AR
255	Primary Congenital Glaucoma	CYP1B1	AR
256	Primary Hyperoxaluria: Type 1	AGXT	AR
257	Primary Hyperoxaluria: Type 2	GRHPR	AR
258	Primary Hyperoxaluria: Type 3	HOGA1	AR
259	Progressive Familial Intrahepatic Cholestasis: Type 2	ABCB11	AR
260	Propionic Acidemia: PCCA Related	PCCA	AR
261	Propionic Acidemia: PCCB Related	PCCB	AR
262	Pseudocholinesterase Deficiency	BCHE	AR
263	Pycnodysostosis	CTSK	AR
264	Pyruvate Carboxylase Deficiency	PC	AR
265	Pyruvate Dehydrogenase Deficiency	PDHB	AR
266	Pyruvate Dehydrogenase Deficiency: X-Linked	PDHA1	XL
267	Renal Tubular Acidosis and Deafness	ATP6V1B1	AR
268	Retinal Dystrophies: RLBP1 Related	RLBP1	AR
269	Retinal Dystrophies: RPE65 Related	RPE65	AR
270	Retinitis Pigmentosa: CERKL Related	CERKL	AR
271	Retinitis Pigmentosa: DHDDS Related	DHDDS	AR
272	Retinitis Pigmentosa: FAM161A Related	FAM161A	AR
273	Rhizomelic Chondrodysplasia Punctata: Type I	PEX7	AR
274	SCID: X-Linked	IL2RG	XL
275	Salla Disease	SLC17A5	AR
276	Sandhoff Disease	HEXB	AR
277	Sanfilippo Syndrome: Type A	SGSH	AR
278	Sanfilippo Syndrome: Type B	NAGLU	AR
279	Sanfilippo Syndrome: Type C	HGSNAT	AR
280	Sanfilippo Syndrome: Type D	GNS	AR
281	Short-Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	AR
282	Sickle-Cell Anemia	HBB	AR
283	Sjogren-Larsson Syndrome	ALDH3A2	AR
284	Sly Syndrome	GUSB	AR
285	Smith-Lemli-Opitz Syndrome	DHCR7	AR
286	Spinal Muscular Atrophy: SMN1 Linked	SMN1	AR
287	Stargardt Disease	ABCA4	AR
288	Stuve-Wiedemann Syndrome	LIFR	AR
289	Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	AR
290	Tay-Sachs Disease	HEXA	AR
291	Trichohepatoenteric Syndrome: Type 1	TTC37	AR
292	Tyrosine Hydroxylase Deficiency	TH	AR
293	Tyrosinemia: Type I	FAH	AR
294	Tyrosinemia: Type II	TAT	AR

#	Diseases	GENES	INHERITANCE
295	Usher Syndrome: Type 1B	MYO7A	AR
296	Usher Syndrome: Type 1C	USH1C	AR
297	Usher Syndrome: Type 1D	CDH23	AR
298	Usher Syndrome: Type 1F	PCDH15	AR
299	Usher Syndrome: Type 2A	USH2A	AR
300	Usher Syndrome: Type 3	CLRN1	AR
301	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	AR
302	Walker-Warburg Syndrome	FKTN	AR
303	Werner Syndrome	WRN	AR
304	Wilson Disease	ATP7B	AR
305	Wiskott-Aldrich Syndrome	WAS	XL
306	Wolcott-Rallison Syndrome	EIF2AK3	AR
307	Wolman Disease	LIPA	AR
308	Xeroderma Pigmentosum: Group A	XPA	AR
309	Xeroderma Pigmentosum: Group C	XPC	AR
310	Zellweger Spectrum Disorders: PEX1 Related	PEX1	AR
311	Zellweger Spectrum Disorders: PEX10 Related	PEX10	AR
312	Zellweger Spectrum Disorders: PEX2 Related	PEX2	AR
313	Zellweger Spectrum Disorders: PEX6 Related	PEX6	AR