GENETIC DISEASE	GENE	MUTATIONS
ACONDROGENESI tipo 1A	TRIP11	Arg253Ter
"	TRIP11	Arg264Ter
"	TRIP11	Gln579Ter
"	TRIP11	Arg1028Ter
"	TRIP11	Gln1160Ter
"	TRIP11	Arg1167Ter
ACONDROGENESI tipo 1B	SLC26A2	Leu132Pro
"	SLC26A2	Arg178Ter
"	SLC26A2	Asn425Asp
"	SLC26A2	Leu483Pro
"	SLC26A2	Gly678Val
ACONDROGENESI II	COL2A1	Gly769Ser
"	COL2A1	Gly691Arg
"	COL2A1	Gly316Asp
"	COL2A1	Gly346Val
ACONDROPLASIA	FGFR3	Gly346Glu
"	FGFR3	Gly375Cys
"	FGFR3	Gly380Arg
"	FGFR3	Gly380Arg
"	FGFR3	Thr394Ser
ALAGILLE	JAG1	Arg184His
"	JAG1	Leu37Ser
"	JAG1	Pro163Leu
"	JAG1	Pro871Arg
"	JAG1	Gly274Asp
"	JAG1	Cys234Tyr
APERT	FGFR2	Ser252Trp
"	FGFR2	Pro253Arg
ATELOSTEOGENESI tipo II	FLNB	Ala173Val
"	FLNB	Met202Val
ATELOSTEOGENESI tipo III	FLNB	Gly751Arg
ATASSIA TELENGIACTESIA	ATM	Val2424Gly
"	ATM	Phe2827Cys
"	ATM	Arg35Ter
"	ATM	Asp2625Glu
"	ATM	Ala2626Pro
"	ATM	Arg2443Ter
BARDET BIEDL	BBS1	Glu549Ter
"	BBS1	Leu518Pro
"	BBS1	Glu234Lys
"	BBS10	Arg34Pro
"	BBS10	Ser311Ala
"	BBS10	Val11Gly
BLOOM	BLM	Cys1036Phe
"	BLM	Trp881Ter
"	BLM	Arg899Ter
"	BLM	Lys272Ter
CANAVAN	ASPA	Tyr231Cys
"	ASPA	Glu285Ala
"	ASPA	Ala305Glu
"	ASPA	Tyr231Cys
CARDIO FACIO CUTANEO	BRAF	Ala246Pro
"	BRAF	Gln257Arg
"	BRAF	Gly469Glu
"	BRAF	Lys499Glu
"	BRAF	Glu501Lys
"	BRAF	Glu501Gly
"	BRAF	Leu485Phe
"	BRAF	Asp638Glu

"	BRAF	Gly534Arg
CONDRODISLASIA PUNTATA tipo 1	PEX7	Leu292Ter
"	PEX7	Ala218Val
"	PEX7	Gly217Arg
"	PEX7	Arg232Ter
COFFIN LOWRY	RPS6KA3	Gly75Val
"	RPS6KA3	Ser227Ala
"	RPS6KA3	Val82Phe
"	RPS6KA3	Arg114Trp
"	RPS6KA3	Gln689Ter
"	RPS6KA3	Arg729Gln
"	RPS6KA3	Arg383Trp
"	RPS6KA3	Ile189Lys
"	RPS6KA3	Phe268Ser
"	RPS6KA3	Thr115Ser
CORNELIA DE LANGE	NIPBL	Tyr2430Cys
"	NIPBL	Arg1723Ter
"	NIPBL	Ala1246Gly
"	NIPBL	Arg1536Ter
"	SMC1A	Glu493Ala
"	SMC1A	Arg496His
"	SMC1A	Ile784Thr
COSTELLO	HRAS	Gly12Ser
"	HRAS	Gly12Ala
"	HRAS	Gly13Asp
"	HRAS	Lys117Arg
"	HRAS	Gly13Cys
"	HRAS	Ala146Thr
"	HRAS	Thr58Ile
"	HRAS	Ala146Val
"	HRAS	Gly12Asp
"	HRAS	Gly12Cys
CROUZON	FGFR2	Cys342Tyr
"	FGFR2	Cys342Arg
"	FGFR2	Cys342Ser
"	FGFR2	Tyr340His
"	FGFR2	Ser354Cys
"	FGFR2	Ala344Ala
"	FGFR2	Ala344Gly
"	FGFR2	Tyr328Cys
"	FGFR2	Ser347Cys
"	FGFR2	Cys342Trp
"	FGFR2	Lys292Glu
"	FGFR2	Trp290Arg
"	FGFR2	Trp290Gly
"	FGFR2	Glu565Ala
"	FGFR2	Ala337Pro
DISAUTONOMIA FAMILIARE	IKBKAP	Arg696Pro
"	IKBKAP	Pro914Leu
DISGENESIA CORTICALE MALFORM.CEREBRALI	TUBB3	ARG262CYS
"	TUBB3	ALA302THR
"	TUBB3	ASP417HIS
"	TUBB3	ASP417ASN
"	TUBB3	GLU410LYS
"	TUBB3	THR178MET
"	TUBB3	GLU205LYS
"	TUBB3	ALA302VAL
"	TUBB3	MET323VAL
DISPLASIA CAMPTOMELICA	SOX9	Tyr440Ter
"	SOX9	Lys173Glu

"	SOX9	His165Tyr
"	SOX9	Ala76Glu
"	SOX9	Phe154Leu
"	SOX9	Ala158Thr
ELHERS-DANLOS	ADAMTS2	Gln225Ter
"	ADAMTS2	Trp795Ter
"	COL1A1	Gly188Asp
"	COL1A1	Gly191Asp
"	COL1A1	Gly197Asp
"	COL1A1	Gly203Cys
"	COL1A1	Gly203Val
"	COL1A1	Gly212Arg
"	COL1A1	Gly266Glu
"	COL1A1	Arg1036Cys
"	COL1A1	Arg1066Cys
"	COL1A1	Arg312Cys
"	COL1A2	Gly109Asp
"	COL1A2	Gly196Val
"	COL3A1	Gly790Ser
"	COL3A1	Gly619Arg
"	COL3A1	Gly883Asp
"	COL3A1	Ala531Thr
"	COL3A1	Gly910Val
"	COL3A1	Gly847Glu
"	COL3A1	Gly1018Asp
"	COL3A1	Gly1006Glu
"	COL3A1	Gly1021Glu
"	COL3A1	Gly136Arg
"	COL3A1	Gly637Ser
"	COL3A1	Gly499Asp
"	COL3A1	Gly793Val
"	COL3A1	Gly415Ser
"	COL3A1	Gly934Glu
"	COL3A1	Gly571Ser
"	COL3A1	Gly16Ser
"	COL3A1	Gly82Asp
"	COL3A1	Gly373Arg
"	COL3A1	Gly385Glu
"	COL3A1	Gly297Arg
"	COL3A1	Gly883Val
"	COL5A1	Leu25Arg
"	COL5A1	Leu25Pro
"	COL5A1	Trp29Ter
"	COL5A1	Gly54Ter
"	COL5A1	Gln127Ter
"	COL5A1	Trp165Ter
"	COL5A1	Thr178Pro
"	COL5A1	Gln222Ter
"	COL5A1	Glu468Ter
"	COL5A1	Gly475Asp
"	COL5A1	Gly493Arg
"	COL5A1	Gly511Val
"	COL5A1	Gly530Ser
"	COL5A1	Gln551Ter
"	COL5A1	Gln552Ter
"	COL5A1	Arg594Ter
"	COL5A1	Gly619Ter
"	COL5A1	Gln729Ter
"	COL5A1	Glu812Asp
"	COL5A1	Glu1013Ter

"	COL5A1	Arg1062Ter
"	COL5A1	Gly1072Glu
"	COL5A1	Gln1110Ter
"	COL5A1	Gly1138Glu
"	COL5A1	Gly1162Arg
"	COL5A1	Gln1190Ter
"	COL5A1	Arg1257Ter
"	COL5A1	Glu1292Lys
"	COL5A1	Gly1393Asp
"	COL5A1	Glu1412Ter
"	COL5A1	Gly1486Cys
"	COL5A1	Gly1489Arg
"	COL5A1	Gly1489Glu
"	COL5A1	Gly1492Ser
"	COL5A1	Gln1518Ter
"	COL5A1	Gln1574Ter
"	COL5A1	Cys1639Ser
"	COL5A1	Cys1639Tyr
"	COL5A1	Tyr1654Ter
"	COL5A1	Gly1832Arg
"	COL5A1	Cys1835Ser
"	COL5A1	Arg792Ter
"	COL5A2	Gly396Arg
"	COL5A2	Gly645Arg
"	COL5A2	Gly1146Ala
"	COL5A2	Gly1149Arg
"	COL5A2	Gly1209Arg
"	PLOD1	Gln49Ter
"	PLOD1	Tyr142Ter
"	PLOD1	Arg319Ter
"	PLOD1	Gln327Ter
"	PLOD1	Trp446Gly
"	PLOD1	Tyr511Ter
"	PLOD1	Trp612Cys
"	PLOD1	Ala667Thr
"	PLOD1	Arg670Ter
"	PLOD1	Gly678Arg
"	PLOD1	His706Arg
"	TNXB	Arg29Trp
"	TNXB	Val1108Met
ELLIS VAN CREVELD	EVC	Arg340Ter
"	EVC	Gln879Ter
"	EVC	Arg443Gln
"	EVC	Ser307Pro
EPIDERMOLISI BOLLOSA	KRT5	Pro25Leu
"	KRT5	Gly550Ala
"	KRT5	Glu477Lys
"	KRT14	Met119Thr
"	KRT14	Asn123Ser
"	KRT14	Arg125Cys
"	KRT14	Arg125His
FENILCHETONURIA	PAH	Arg261Ter
"	PAH	Arg408Gln
"	PAH	Phe299Cys
"	PAH	Ser349Pro
"	PAH	Ala322Gly
"	PAH	Asp415Asn
"	PAH	Ile306Val
"	PAH	Val388Met
"	PAH	Pro244Leu

"	PAH	Met1Ile
"	PAH	Leu333Phe
"	PAH	Glu390Gly
"	PAH	Ser359Ter
"	PAH	Leu98Ser
"	PAH	Thr380Met
"	PAH	Gly46Ser
"	PAH	Ala47Val
"	PAH	Ser87Arg
"	PAH	Arg176Leu
"	PAH	Val245Ala
"	PAH	Pro407Leu
"	PAH	Ile65Thr
"	PAH	Glu76Gly
FIBROSI CISTICA	CFTR	Ser18Arg
"	CFTR	Arg31Cys
"	CFTR	Gln39Ter
"	CFTR	Ala46Asp
"	CFTR	Glu56Lys
"	CFTR	Arg59Lys
"	CFTR	Glu60Ter
"	CFTR	Pro67Leu
"	CFTR	Arg74Trp
"	CFTR	Arg75Ter
"	CFTR	Arg75Gln
"	CFTR	Gly85Glu
"	CFTR	Leu88Ile
"	CFTR	Glu92Lys
"	CFTR	Glu92Ter
"	CFTR	Gln98Ter
"	CFTR	Ile105Ser
"	CFTR	Tyr109Gly
"	CFTR	Asp110His
"	CFTR	Arg117Cys
"	CFTR	Arg117His
"	CFTR	Tyr122Ter
"	CFTR	Ile148Leu
"	CFTR	Ile148Thr
"	CFTR	Gly178Glu
"	CFTR	Leu183Phe
"	CFTR	His199Tyr
"	CFTR	Pro205Ser
"	CFTR	Leu206Trp
"	CFTR	Gln220Ter
"	CFTR	Leu227Arg
"	CFTR	Gly241Glu
"	CFTR	Cys276Ter
"	CFTR	Phe316Leu
"	CFTR	Gly330Ter
"	CFTR	Arg334Trp
"	CFTR	Ile336Lys
"	CFTR	Thr338Ile
"	CFTR	Ser341Pro
"	CFTR	Phe342His
"	CFTR	Arg347His
"	CFTR	Arg347Pro
"	CFTR	Arg352Gln
"	CFTR	Gln359Lys
"	CFTR	Trp361Gly
"	CFTR	Gln378Ala

"	CFTR	Asn386Ile
"	CFTR	Trp401Ter
"	CFTR	Gln414Ter
"	CFTR	Ile444Arg
"	CFTR	Lys447Arg
"	CFTR	Ala455Glu
"	CFTR	Ser466Ter
"	CFTR	Leu467Pro
"	CFTR	Gly473Glu
"	CFTR	Ser489Ter
"	CFTR	Ser492Phe
"	CFTR	Gln493Ter
"	CFTR	Ile507del
"	CFTR	Phe508del
"	CFTR	Tyr515Ter
"	CFTR	Val520Phe
"	CFTR	Gln525Ter
"	CFTR	Gly542Ter
"	CFTR	Ser549Arg
"	CFTR	pSer549Asn
"	CFTR	Gly551Val
"	CFTR	Gly551Ser
"	CFTR	Gly551Asp
"	CFTR	Gln552Ter
"	CFTR	Arg553Ter
"	CFTR	Leu558Ser
"	CFTR	Ala559Thr
"	CFTR	Arg560Lys
"	CFTR	Arg560Thr
"	CFTR	Ala561Glu
"	CFTR	Asp565Met
"	CFTR	Tyr569Asp
"	CFTR	Gly576Ala
"	CFTR	Asp579Gly
"	CFTR	Glu585Ter
"	CFTR	Asp614Gly
"	CFTR	Ser641Arg
"	CFTR	Arg658Lys
"	CFTR	Thr663Arg
"	CFTR	Arg668Cys
"	CFTR	Leu671Ter
"	CFTR	Lys684Ser
"	CFTR	Gln685Thr
"	CFTR	Lys684Asn
"	CFTR	Arg709Ter
"	CFTR	Lys710Ter
"	CFTR	Glu726Arg
"	CFTR	Leu732Ter
"	CFTR	Val739Tyr
"	CFTR	Val754Met
"	CFTR	Arg764Ter
"	CFTR	Arg785Ter
"	CFTR	Arg792Ter
"	CFTR	Ser809Ile
"	CFTR	Leu818Trp
"	CFTR	Ser821Arg
"	CFTR	Glu822Ter
"	CFTR	Glu831Ter
"	CFTR	Trp846Ter
"	CFTR	Tyr849Ter

"	CFTR	Arg851Ter
"	CFTR	Phe861Leu
"	CFTR	Gln890Ter
"	CFTR	Ser912Ter
"	CFTR	Val922Glu
"	CFTR	Leu927Pro
"	CFTR	Ser945Leu
"	CFTR	Ala959His
"	CFTR	Gly970Arg
"	CFTR	Ser977Phe
"	CFTR	Leu997Phe
"	CFTR	Val1001Asp
"	CFTR	Ile1027Thr
"	CFTR	Phe1052Val
"	CFTR	His1054Asp
"	CFTR	Gly1061Arg
"	CFTR	Leu1065Pro
"	CFTR	Arg1066Cys
"	CFTR	Arg1066His
"	CFTR	Gly1069Arg
"	CFTR	Arg1070Trp
"	CFTR	Arg1070Gln
"	CFTR	Phe1074Leu
"	CFTR	Leu1077Pro
"	CFTR	Trp1089Ter
"	CFTR	Tyr1092Ter
"	CFTR	Met1101Lys
"	CFTR	Glu1104Ter
"	CFTR	Asp1152His
"	CFTR	Arg1158Ter
"	CFTR	Arg1162Ter
"	CFTR	Arg1162Leu
"	CFTR	Lys1177Ser
"	CFTR	Thr1179Ile
"	CFTR	Ser1196Ter
"	CFTR	Asp1202Ala
"	CFTR	Trp1204Ter
"	CFTR	Thr1220Lys
"	CFTR	Ser1231Pro
"	CFTR	Ile1234Val
"	CFTR	Ser1235Arg
"	CFTR	Gly1244Glu
"	CFTR	Lys1250Arg
"	CFTR	Ser1251Asn
"	CFTR	Ser1255Pro
"	CFTR	Ser1255Ter
"	CFTR	Leu1258Phe
"	CFTR	Asp1270Asn
"	CFTR	Trp1282Ter
"	CFTR	Ile1295Phe
"	CFTR	Ser1297Phe
"	CFTR	Asn1303Lys
"	CFTR	Gln1313Ter
"	CFTR	Gly1349Asp
"	CFTR	Glu1371Ter
"	CFTR	Cys1400Ter
"	CFTR	Gln1412Ter
"	CFTR	Glu1418Arg
"	CFTR	Ser1435Gly
GALATTOSEMIA	GALT	Arg333Trp

"	GALT	Val44Met
"	GALT	Asn314Asp
"	GALT	Gln188Arg
"	GALT	Leu74Pro
"	GALT	Phe171Ser
"	GALT	His319Gln
"	GALT	Ser135Leu
"	GALT	Pro183Thr
"	GALT	Leu218Leu
"	GALT	Lys285Asn
"	GALT	Glu203Lys
"	GALT	Arg333Gly
"	GALT	Phe194Leu
"	GALT	Asn314Asp
"	GALT	Leu29Ala
GAUCHER	GALT	Arg159Gln
"	GALT	Arg159Trp
"	GALT	Arg170Leu
"	GALT	Asn227Ser
"	GALT	Ser235Pro
"	GALT	Phe252Ile
"	GALT	His294Gln
"	GALT	Arg296Gln
"	GALT	Gly416Ser
"	GALT	Asn409Ser
"	GALT	Leu483Pro
"	GALT	Val433Leu
"	GALT	Asp448His
"	GALT	Asp448Val
"	GALT	Arg502Cys
"	GALT	Arg502His
"	GALT	Arg535His
"	GALT	Glu69Ter
HOLT ORAM	TBX5	Arg237Gln
"	TBX5	Gly80Arg
"	TBX5	Arg237Trp
"	TBX5	Gln49Lys
"	TBX5	Ile54Thr
"	TBX5	Tyr136Ter
"	TBX5	Ser84Leu
IPOCONDROPLASIA	FGFR3	Gln115Leu
"	FGFR3	Arg200Cys
"	FGFR3	Asn262His
"	FGFR3	Thr264Met
"	FGFR3	Gly268Cys
"	FGFR3	Tyr278Cys
"	FGFR3	Gly295Cys
"	FGFR3	Leu324Val
"	FGFR3	Asn328Ile
"	FGFR3	Gly342Cys
"	FGFR3	Ser351Cys
"	FGFR3	Glu360Lys
"	FGFR3	Val381Glu
"	FGFR3	Gln485Arg
"	FGFR3	Ile538Val
"	FGFR3	Asn540Lys
"	FGFR3	Asn540Lys
"	FGFR3	Asn540Ser
"	FGFR3	Asn540Thr
"	FGFR3	Lys650Asn

"	FGFR3	Lys650Asn
"	FGFR3	Lys650Gln
"	FGFR3	Ala162Thr
IPOFOSFATASIA	ALPL	Arg54Cys
"	ALPL	Arg54Pro
"	ALPL	Gln190Pro
"	ALPL	Ala16Val
"	ALPL	Tyr419His
"	ALPL	Glu174Lys
"	ALPL	Arg435Ter
LISSENCEFALIA X-LINKED	DCX	ARG192TRP
"	DCX	ASP62ASN
"	DCX	TYR125HIS
"	DCX	ARG59LEU
"	DCX	THR203ARG
LISSENCEFALIA tipo 3	TUBA1A	ARG402HIS
"	TUBA1A	ILE188LEU
"	TUBA1A	PRO263THR
"	TUBA1A	SER419LEU
"	TUBA1A	LEU397PRO
"	TUBA1A	ARG422CYS
"	TUBA1A	ARG422HIS
"	TUBA1A	ILE5LEU
JOUBERT sindrom.3	AHI1	Val443Asp
"	AHI1	Arg589Ter
"	AHI1	Arg723Gln
"	AHI1	Arg329Ter
"	AHI1	Arg495His
"	AHI1	Arg351Leu
"	AHI1	Trp7Cys
JOUBERT sindrom.5	CEP290	Leu750Ter
"	CEP290	Gln44Ter
JOUBERT sindrom.6	TMEM67	Asn90Lys
"	TMEM67	Asp110Gly
"	TMEM67	Glu124Lys
"	TMEM67	Arg213Cys
"	TMEM67	Asp301Glu
"	TMEM67	Lys329Thr
"	TMEM67	Arg79Gln
JOUBERT sindrom.8	ALR13B	Trp82Ter
"	ALR13B	Arg200Cys
"	ALR13B	Lys233Ter
JOUBERT sindrom.7	RPGRIP1L	Thr615Pro
"	RPGRIP1L	Gln253Ter
"	RPGRIP1L	Ala695Pro
"	RPGRIP1L	Gln684Ter
"	RPGRIP1L	Pro1122Ser
JOUBERT sindrom.9	CC2D2A	Arg1528Cys
"	CC2D2A	Arg950Ter
"	CC2D2A	Arg1049Ter
"	CC2D2A	Thr1116Met
"	CC2D2A	Glu1447Ala
"	CC2D2A	Arg73Leu
JOUBERT sindrom.2	TMEM216	Arg73His
KABUKI	KMT2D	Arg5179His
"	KMT2D	Lys4527Ter
"	KMT2D	Arg5454Ter
"	KMT2D	Thr5464Met
MARFAN	FBN1	Arg1137Pro
"	FBN1	Cys2307Ser

"	FBN1	Trp2756Ter
"	FBN1	Cys1249Ser
"	FBN1	Cys1663Arg
"	FBN1	Cys2221Ser
"	FBN1	Tyr2113Ter
"	FBN1	Asn2144Ser
"	FBN1	Asn548Ile
"	FBN1	Asp723Ala
"	FBN1	Cys1074Arg
"	FBN1	Arg2776Ter
"	FBN1	Arg122Cys
"	FBN1	Gly1127Ser
"	FBN1	Cys1223Tyr
"	FBN1	Cys1117Tyr
"	FBN1	Cys1242Tyr
"	FBN1	Lys1043Arg
"	FBN1	Asn1131Tyr
"	FBN1	Cys1265Arg
"	FBN1	Arg1170His
"	FBN1	Arg529Ter
"	FBN1	Gly985Glu
"	FBN1	Gly1013Arg
"	FBN1	Glu1073Lys
"	FBN1	Tyr754Cys
"	FBN1	Arg240Cys
"	FBN1	Cys1032Tyr
"	FBN1	Cys1129Tyr
"	FBN1	Cys1221Tyr
"	FBN1	Cys1086Tyr
"	FBN1	Arg485Cys
"	FBN1	Arg2576Cys
MECKEL	MKS1	Arg158Ter
"	MKS1	Arg166Trp
"	MKS1	Glu277Ter
"	MKS1	Gln350Ter
MICROCEFALIA	ASPM	Gln3060Ter
"	ASPM	Trp1326Ter
"	ASPM	Arg117Ter
"	ASPM	Tyr2063Ter
"	ASPM	Arg797Ter
MUCOLIPIDOSI	MCOLN1	Arg321Ter
"	MCOLN1	Asp362Tyr
"	MCOLN1	Arg102Ter
"	MCOLN1	Arg403Cys
NAIL-PATELLA	LMX1B	Arg198Ter
"	LMX1B	Cys95Phe
"	LMX1B	Gln59Ter
"	LMX1B	Arg208Ter
"	LMX1B	Arg200Gln
"	LMX1B	Arg226Ter
NOONAN	PTPN11	Ala72Ser
"	PTPN11	Ala72Gly
"	PTPN11	Asn308Asp
"	PTPN11	Asn308Ser
"	PTPN11	Tyr279Cys
"	PTPN11	Thr468Met
"	PTPN11	Ser502Thr
"	PTPN11	Tyr63Cys
"	PTPN11	Tyr62Asp
"	PTPN11	Asp61Gly

"	PTPN11	Thr73Ile
"	PTPN11	Phe285Ser
"	PTPN11	Gln79Arg
"	PTPN11	Thr411Met
"	PTPN11	Ala461Thr
"	PTPN11	Gly464Ala
"	PTPN11	Gln510Pro
"	PTPN11	Gln510Arg
"	PTPN11	Arg138Ter
"	PTPN11	Thr2Ile
"	SOS1	Thr266Lys
"	SOS1	Met269Arg
"	SOS1	Arg552Gly
"	SOS1	Arg552Ser
"	SOS1	Trp432Arg
"	KRAS	Thr58Ile
"	KRAS	Val14Ile
"	KRAS	Val152Gly
"	KRAS	Asp153Val
"	KRAS	Lys5Glu
"	KRAS	Gly60Ser
"	RAF1	Ser257Leu
"	RAF1	Pro261Ser
"	RAF1	Thr491Arg
"	RAF1	Leu613Val
"	BRAF	Thr241Met
"	BRAF	Thr241Arg
"	BRAF	Trp531Cys
"	BRAF	Leu597Val
"	NRAS	Thr50Ile
"	NRAS	Gly60Glu
"	CBL	Gln367Pro
"	CBL	Lys382Glu
"	CBL	Asp390Tyr
"	CBL	Arg420Gln
"	CBL	Tyr371His
"	CBL	Cys384Arg
"	CBL	Cys396Arg
"	CBL	Tyr371Cys
OLOPROSENCEFALIA	**SHH**	Gly31Arg
"	**SHH**	Gln100Ter
"	**SHH**	Lys105Ter
"	**SHH**	Trp117Gly
"	**SHH**	Trp117Arg
"	**SHH**	Val224Glu
"	**SHH**	Ala226Thr
"	**SHH**	Glu284Ter
"	**SHH**	Ala384Thr
"	**SHH**	Gly290Asp
"	**SHH**	Pro424Ala
"	**SHH**	Asp88Val
"	**SHH**	Glu256Ter
"	**SHH**	Trp128Ter
"	**SHH**	Asn115Lys
"	SIX3	Leu226Val
"	SIX3	Arg257Pro
"	SIX3	Val250Ala
"	SIX3	Gly69Asp
"	SIX3	Trp113Cys
"	SIX3	Glu129Ter

"	SIX3	Gly37Cys
OSTEOGENESI IMPERFETTA	COL1A1	Gly85Arg
"	COL1A1	Gly1017Cys
"	COL1A1	Gly1017Cys
OSTEOGENESI IMPERFETTA, TIPO I	COL1A1	Gly94Cys
"	COL1A1	Gly178Cys
"	COL1A1	Arg963Ter
"	COL1A1	Arg134Cys
"	COL1A1	Gly901Ser
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly391Arg
"	COL1A1	Gly97Asp
"	COL1A1	Gly541Asp
"	COL1A1	Gly559Asp
"	COL1A1	Gly673Asp
"	COL1A1	Gly667Arg
"	COL1A1	Gly691Cys
"	COL1A1	Gly718Cys
"	COL1A1	Gly847Arg
"	COL1A1	Gly883Asp
"	COL1A1	Gly904Cys
"	COL1A1	Gly913Ser
"	COL1A1	Gly988Cys
"	COL1A1	Gly1009Ser
"	COL1A1	Gly1003Ser
"	COL1A1	Gly637Val
"	COL1A1	Gly565Val
"	COL1A1	Gly355Asp
OSTEOGENESIS IMPERFECTA, TYPE IIC	COL1A1	Gly1006Val
OSTEOGENESIS IMPERFECTA, TYPE IIA	COL1A1	Gly973Val
OSTEOGENESIS IMPERFECTA, TYPE IIA	COL1A1	Gly256Val
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Gly802Val
"	COL1A1	Gly415Ser
"	COL1A2	Gly976Asp
"	COL1A2	Gly805Asp
"	COL1A2	Gly259Cys
"	COL1A2	Gly472Cys
"	COL1A2	Gly421Asp
"	COL1A1	Gly586Val
"	COL1A2	Gly694Arg
"	COL1A2	Gly580Asp
"	COL1A1	Ala1387Val
"	COL1A2	Gly865Ser
"	COL1A2	Gly907Asp
"	COL1A2	Gly502Ser
"	COL1A2	Gly547Asp
"	COL1A1	Trp94Cys
"	COL1A1	Gly748Cys
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly862Ser
"	COL1A1	Gly661Ser
"	COL1A1	Gly526Cys
"	COL1A1	Gly154Arg
"	COL1A1	Gly844Ser
OSTEOGENESIS IMPERFECTA, TYPE III/IV	COL1A1	Gly415Cys
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly352Ser
"	COL1A2	Gly1006Ala
"	COL1A2	Gly586Val
"	COL1A2	Gly751Ser
"	COL1A2	Gly277Tyr
"	COL1A1	Gly76Glu
"	COL1A2	Gly859Ser

"	COL1A2	Gly1090Asp
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A1	Gly832Ser
"	COL1A1	Gly175Cys
"	COL1A2	Gly646Cys
"	COL1A2	Gly586Val
"	COL1A2	Gly1012Arg
"	COL1A2	Gly379Ala
OSTEOGENESIS IMPERFECTA, TYPE VII	CRTAP	Gln276Ter
"	CRTAP	Tyr187Ter
OSTEOGENESI IMPERFETTA	LEPRE1	Tyr552Ter
"	LEPRE1	Arg368Ter
POLIMICROGIRIA,IPOPLASIA NERVO OTTICO	TUBA8	14-BP DEL
POLIMICROGIRIA	TUBB2B	SER172PRO
"	TUBB2B	LEU228PRO
"	TUBB2B	PHE265LEU
"	TUBB2B	ASP417ASN
"	TUBB2B	ASN256SER
"	TUBB2B	LEU117PRO
"	TUBB2B	GLU421LYS
RENE POLICISTICO DELL'ADULTO	PKD1	Gln1273Ter
"	PKD1	Arg4227Ter
"	PKD1	Gln3837Ter
"	PKD1	Cys4086Ter
"	PKD1	Tyr3818Ter
"	PKD1	Arg324Leu
"	PKD1	Leu845Ser
"	PKD1	Gln1922Ter
"	PKD1	Trp4139Ter
"	PKD2	Trp380Ter
"	PKD2	Arg742Ter
"	PKD2	Gln405Ter
"	PKD2	Arg464Ter
"	PKD2	Asp511Val
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Thr36Met
"	PKHD1	Ser1664Phe
"	PKHD1	Ser3018Phe
"	PKHD1	Val1741Met
"	PKHD1	Arg2671Ter
"	PKHD1	Ile3553Thr
"	PKHD1	Arg496Ter
"	PKHD1	Val3471Gly
PFEIFFER	FGFR1	Pro252Arg
"	FGFR1	Cys342Tyr
"	FGFR1	Cys342Arg
"	FGFR1	Thr341Pro
"	FGFR1	Ser252Phe
"	FGFR1	Pro253Ser
"	FGFR1	Trp290Cys
"	FGFR1	Ser351Cys
"	FGFR1	Ser267Pro
"	FGFR1	Trp290Cys
"	FGFR1	Glu565Ala
"	FGFR1	Asp321Ala
RETT	MECP2	Arg133Cys
"	MECP2	Phe155Ser
"	MECP2	Arg270Ter
"	MECP2	Thr158Met
"	MECP2	Arg106Trp
"	MECP2	Arg294Ter
"	MECP2	Arg306Cys

"	MECP2	Arg168Ter
"	MECP2	Arg255Ter
"	MECP2	Glu455Ter
"	MECP2	Leu100Val
"	MECP2	Pro152Ala
"	MECP2	Ala2Val
SAETHRE-CHOTZEN	TWIST1	Tyr103Ter
"	TWIST1	Gln119Pro
"	TWIST1	Tyr107Ter
"	TWIST1	Ser127Ter
"	TWIST1	Glu130Ter
"	TWIST1	Leu135Pro
"	TWIST1	Glu181Ter
"	TWIST1	Ile156Val
"	TWIST1	Gln28Ter
SECKEL	ATR	Asp1879Tyr
"	ATR	Met1159Ile
SMITH-LEMLI-OPITZ	DHCR7	His119Leu
"	DHCR7	Gly244Arg
"	DHCR7	Trp248Cys
"	DHCR7	Thr93Met
"	DHCR7	Trp151Ter
"	DHCR7	Val326Leu
"	DHCR7	Trp37Ter
"	DHCR7	Arg352Trp
"	DHCR7	Arg404Cys
"	DHCR7	Thr289Ile
"	DHCR7	Tyr280Cys
"	DHCR7	Met1Leu
"	DHCR7	Glu448Lys
"	DHCR7	Phe284Leu
"	DHCR7	Met1Val
"	DHCR7	Arg352Gln
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	MET34THR
	GJB2	TRP24TER
	GJB2	TRP77ARG
	GJB2	1-BP DEL, 35G
	GJB2	GLU47TER
	GJB2	GLU118DEL
	GJB2	ARG184PRO
	GJB2	ARG143TRP
	GJB2	1-BP DEL, 167T
	GJB2	LEU90PRO
	GJB2	VAL37ILE
	GJB2	ASP159VAL
	GJB2	VAL84LEU
	GJB2	VAL84MET
SORDITA' CONGENITA AUT.DOMINANTE	GJB6	THR5MET
SOTOS	NSD1	Ser437Ter
"	NSD1	His2143Glu
"	NSD1	Cys2183Ser
"	NSD1	Arg1320Ter
"	NSD1	Cys2202Tyr
TAY-SACHS DISEASE	HEXA	Glu482Lys
"	HEXA	Arg178Cys
"	HEXA	Arg170Gln
"	HEXA	Trp420Cys
"	HEXA	Gly250Asp
"	HEXA	Ser210Phe
"	HEXA	Arg137Ter

"	HEXA	Arg393Ter
"	HEXA	Trp26Ter
"	HEXA	Arg178Leu
"	HEXA	Met1Val
"	HEXA	Trp485Arg
"	HEXA	Tyr180Ter
"	HEXA	Asp258His
"	HEXA	Arg170Trp
"	HEXA	Phe211Ser
"	HEXA	Leu127Arg
"	HEXA	His204Arg
"	HEXA	Met301Arg
"	HEXA	Gly454Ser
"	HEXA	Leu39Arg
"	HEXA	Trp392Ter
"	HEXA	Leu451Val
TIROSINEMIA	FAH	Asn16Ile
"	FAH	Ala134Asp
"	FAH	Glu357Ter
"	FAH	Glu364Ter
"	FAH	Arg341Trp
"	FAH	Glu337Ser
"	FAH	Arg381Gly
"	FAH	Trp262Ter
"	FAH	Gln279Arg
TREACHER COLLINS	TCOF1	Gln252Ter
"	TCOF1	Tyr50Cys
"	TCOF1	Arg911Ter
WILSON	ATP7B	His714Gln
"	ATP7B	Asn915Ser
"	ATP7B	His1069Gln
"	ATP7B	Gly1267Arg
"	ATP7B	Arg778Leu
"	ATP7B	Asp765Asn
"	ATP7B	Gly943Ser
"	ATP7B	Arg919Gly
"	ATP7B	Ala874Val
"	ATP7B	Asn1270Ser
"	ATP7B	Arg969Gln
"	ATP7B	Thr766Arg
"	ATP7B	Met645Arg
"	ATP7B	Gly1176Arg
"	ATP7B	Ile1148Thr
"	ATP7B	Gln289Ter
"	ATP7B	Leu708Pro
"	ATP7B	Gly691Arg
"	ATP7B	Ile1148Thr
ZELLWEGER	PEX1	Leu664Pro