

List disease genes and mutations	GENE	MUTAZIONI
ACHONDROGENESIS, TYPE IA	TRIP11	Arg264Ter
ACHONDROGENESIS, TYPE IA	TRIP11	Trp1224Ter
ACHONDROGENESIS, TYPE IA	TRIP11	Asn701Ser
ACHONDROGENESIS, TYPE IB	SLC26A2	Arg178Ter
ACHONDROGENESIS, TYPE IB	SLC26A2	Asn425Asp
ACHONDROGENESIS, TYPE IB	SLC26A2	Gly678Val
ACHONDROGENESIS, TYPE IB	SLC26A2	Val340del
HYPOCHONDROGENESIS	COL2A1	Gly943Ser
HYPOCHONDROGENESIS	COL2A1	Gly574Ser
HYPOCHONDROGENESIS	COL2A1	Gly853Glu
ACHONDROGENESIS, TYPE II	COL2A1	Gly769Ser
ACHONDROGENESIS, TYPE II	COL2A1	Gly691Arg
ACHONDROGENESIS, TYPE II	COL2A1	Gly316Asp
ACHONDROGENESIS, TYPE II	COL2A1	Gly346Val
ACHONDROPLASIA	FGFR3	Gly380Arg
ACHONDROPLASIA	FGFR3	Gly375Cys
ACHONDROPLASIA	FGFR3	Leu377Arg
ACHONDROPLASIA	FGFR3	Ser279Cys
ALAGILLE SYNDROME 1	JAG1	Arg184Cys
ALAGILLE SYNDROME 1	JAG1	Arg184His
TETRALOGY OF FALLOT	JAG1	Gly274Asp
ALAGILLE SYNDROME 1	JAG1	Leu37Ser
APERT SYNDROME	FGFR2	Ser252Trp
APERT SYNDROME	FGFR2	Pro253Arg
APERT SYNDROME	FGFR2	Ser252Phe
ATELOSTEOGENESIS, TYPE I	FLNB	Ala173Val
ATELOSTEOGENESIS, TYPE I	FLNB	Met202Val
ATELOSTEOGENESIS, TYPE III	FLNB	Gly751Arg
ATELOSTEOGENESIS, TYPE I	FLNB	Ala173Thr
ATAXIA-TELANGIECTASIA VARIANT	ATM	Val2424Gly
ATAXIA-TELANGIECTASIA VARIANT	ATM	Phe2827Cys
ATAXIA-TELANGIECTASIA	ATM	Arg35Ter
ATAXIA-TELANGIECTASIA WITHOUT IMMUNODEFICIENCY	ATM	Leu2656Pro
ATAXIA-TELANGIECTASIA	ATM	Asp2625Glu
ATAXIA-TELANGIECTASIA	ATM	Ala2626Pro
ATAXIA-TELANGIECTASIA VARIANT	ATM	Tyr2677Cys
ATAXIA-TELANGIECTASIA VARIANT	ATM	Ala2067Asp
BARDET-BIEDL SYNDROME 1	BBS1	Met390Arg
BARDET-BIEDL SYNDROME 1	BBS1	Glu549Ter
BARDET-BIEDL SYNDROME 1	BBS1	Leu518Pro
BARDET-BIEDL SYNDROME 1/7, DIGENIC	BBS1	Glu234Lys
BARDET-BIEDL SYNDROME 10	BBS10	Arg34Pro
BARDET-BIEDL SYNDROME 10	BBS10	Ser311Ala
BARDET-BIEDL SYNDROME 10	BBS10	Val11Gly
BARDET-BIEDL SYNDROME 10	BBS10	Cys91Trp
BLOOM	BLM	Cys1036Phe
BLOOM	BLM	Trp881Ter
BLOOM	BLM	Arg899Ter
BLOOM	BLM	Lys272Ter
CANAVAN DISEASE	ASPA	Glu285Ala
CANAVAN DISEASE	ASPA	Cys152Arg
CANAVAN DISEASE	ASPA	Ala305Glu
CANAVAN DISEASE	ASPA	Cys218Ter
CANAVAN DISEASE	ASPA	Tyr231Ter
CANAVAN DISEASE	ASPA	Tyr231Cys
CANAVAN DISEASE	ASPA	Glu24Gly
CANAVAN DISEASE	ASPA	Asp249Val
CANAVAN DISEASE, MILD	ASPA	Arg71His
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Ala246Pro
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Gln257Arg
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Leu485Phe
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Glu501Lys
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Glu501Gly
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Gly534Arg
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Asp638Glu
CARDIOFACIOCUTANEOUS SYNDROME 1	BRAF	Thr241Pro
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1	PEX7	Leu292Ter
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1	PEX7	Ala218Val
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1	PEX7	Gly217Arg
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1	PEX7	Arg232Ter
COFFIN-LOWRY SYNDROME	RPS6KA3	Gly75Val
COFFIN-LOWRY SYNDROME	RPS6KA3	Ser227Ala
COFFIN-LOWRY SYNDROME	RPS6KA3	Val82Phe
COFFIN-LOWRY SYNDROME	RPS6KA3	Arg114Trp
COFFIN-LOWRY SYNDROME	RPS6KA3	Gln689Ter

COFFIN-LOWRY SYNDROME	RPS6KA3	Arg729Gln
COFFIN-LOWRY SYNDROME, MILD	RPS6KA3	Ile189Lys
COFFIN-LOWRY SYNDROME	RPS6KA3	Phe268Ser
CORNELIA DE LANGE SYNDROME 1	NIPBL	Met1Lys
CORNELIA DE LANGE SYNDROME 1	NIPBL	Tyr2430Cys
CORNELIA DE LANGE SYNDROME 1	NIPBL	Ile1206del
CORNELIA DE LANGE SYNDROME 1	NIPBL	Arg1723Ter
CORNELIA DE LANGE SYNDROME 1	NIPBL	Arg1536Ter
CORNELIA DE LANGE SYNDROME 2	SMC1A	Glu493Ala
CORNELIA DE LANGE SYNDROME 2	SMC1A	Arg496His
CORNELIA DE LANGE SYNDROME 2	SMC1A	Ile784Thr
COSTELLO SYNDROME	HRAS	Gly12Ser
COSTELLO SYNDROME	HRAS	Gly12Ala
COSTELLO SYNDROME	HRAS	Gly13Asp
COSTELLO SYNDROME	HRAS	Lys117Arg
COSTELLO SYNDROME	HRAS	Gly13Cys
COSTELLO SYNDROME	HRAS	Ala146Thr
COSTELLO SYNDROME	HRAS	Thr58Ile
COSTELLO SYNDROME	HRAS	Ala146Val
COSTELLO SYNDROME, SEVERE	HRAS	Gly12Asp
COSTELLO SYNDROME	HRAS	Gly12Cys
CROUZON SYNDROME	FGFR2	Cys342Tyr
CROUZON SYNDROME	FGFR2	Cys342Arg
CROUZON SYNDROME	FGFR2	Cys342Ser
CROUZON SYNDROME	FGFR2	Tyr340His
CROUZON SYNDROME	FGFR2	Ser354Cys
CROUZON SYNDROME	FGFR2	Ala344Ala
CROUZON SYNDROME	FGFR2	Tyr328Cys
CROUZON SYNDROME	FGFR2	Ser347Cys
CROUZON SYNDROME	FGFR2	Cys342Trp
CROUZON SYNDROME	FGFR2	Ala337Pro
DYSAUTONOMIA, FAMILIAL	IKBKAP	Arg696Pro
DYSAUTONOMIA, FAMILIAL	IKBKAP	Pro914Leu
CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFO	TUBB3	Thr178Met
CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFOR	TUBB3	Glu205Lys
CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFOR	TUBB3	Ala302Val
CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFOR	TUBB3	Met323Val
CAMPOMELIC DYSPLASIA WITH AUTOSOMAL SEX REVERSAL	SOX9	Tyr440Ter
ACAMPOMELIC CAMPOMELIC DYSPLASIA	SOX9	Lys173Glu
ACAMPOMELIC CAMPOMELIC DYSPLASIA	SOX9	His165Tyr
ACAMPOMELIC CAMPOMELIC DYSPLASIA	SOX9	Ala76Glu
CAMPOMELIC DYSPLASIA	SOX9	Phe154Leu
CAMPOMELIC DYSPLASIA WITH AUTOSOMAL SEX REVERSAL	SOX9	Ala158Thr
EHLERS-DANLOS SYNDROME, TYPE VIIC	ADAMTS2	Gln225Ter
EHLERS-DANLOS SYNDROME, TYPE VIIC	ADAMTS2	Trp795Ter
EHLERS-DANLOS SYNDROME, AUTOSOMAL RECESSIVE, CARD	COL1A2	Glu1201Ter
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly790Ser
EHLERS-DANLOS SYNDROME, TYPE IV, VARIANT	COL3A1	Gly619Arg
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly883Asp
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly910Val
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly847Glu
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly1018Asp
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly1006Glu
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly1021Glu
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly136Arg
EHLERS-DANLOS SYNDROME, TYPE III	COL3A1	Gly637Ser
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly499Asp
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly793Val
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly415Ser
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly934Glu
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly571Ser
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly16Ser
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly82Asp
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly373Arg
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly385Glu
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly297Arg
EHLERS-DANLOS SYNDROME, TYPE IV	COL3A1	Gly883Val
EHLERS-DANLOS SYNDROME, TYPE I	COL5A1	Cys1181Ser
EHLERS-DANLOS SYNDROME, TYPE I	COL5A1	Gly1489Glu
EHLERS-DANLOS SYNDROME, TYPE I	COL5A1	Arg792Ter
EHLERS-DANLOS SYNDROME, TYPE I	COL5A1	Arg1062Ter
EHLERS-DANLOS SYNDROME, TYPE II	COL5A2	Gly934Arg
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Arg319Ter
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Gly678Arg
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Glu532del
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Tyr511Ter
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Trp612Cys
EHLERS-DANLOS SYNDROME, TYPE VI	PLOD1	Arg670Ter

EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	TNXB	Val1195Met
EHLERS-DANLOS-LIKE SYNDROME DUE TO TENASCIN-X DEFICIT	TNXB	Arg4072Cys
ELLIS-VAN CREVELD SYNDROME	EVC	Gln879Ter
ELLIS-VAN CREVELD SYNDROME	EVC	Arg340Ter
ELLIS-VAN CREVELD SYNDROME	EVC	Arg443Gln
ELLIS-VAN CREVELD SYNDROME	EVC	Ser307Pro
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT5	Glu475Gly
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT5	Leu462Pro
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT5	Ile161Ser
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT5	Met327Thr
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT5	Asn329Lys
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT5	Lys173Asn
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT5	Asn193Lys
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT5	Leu174Phe
EPIDERMOLYSIS BULLOSA SIMPLEX WITH MOTTLED PIGMENTA	KRT5	Pro25Leu
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT5	Val7Ala
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT5	Ser181Pro
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT5	Val186Leu
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE, M	KRT5	Glu477Ter
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED, WITH SEVEI	KRT5	Lys472Ter
EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1	KRT5	Glu170Lys
EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1	KRT5	Glu418Lys
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT14	Leu384Pro
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Arg125Cys
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Arg125His
EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1	KRT14	Glu144Ala
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT14	Glu375del
EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1	KRT14	Tyr204Ter
EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED	KRT14	Met272Arg
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Met119Thr
EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED	KRT14	Met119Ile
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Tyr415His
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Leu419Gln
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Glu422Lys
EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA TYPE	KRT14	Asn123Ser
PHENYLKETONURIA	PAH	Arg408Trp
PHENYLKETONURIA	PAH	Leu311Pro
PHENYLKETONURIA	PAH	Glu280Lys
PHENYLKETONURIA	PAH	Arg111Ter
PHENYLKETONURIA	PAH	Arg261Gln
PHENYLKETONURIA	PAH	Arg252Trp
PHENYLKETONURIA	PAH	Met1Val
PHENYLKETONURIA	PAH	Arg158Gln
PHENYLKETONURIA	PAH	Arg243Ter
PHENYLKETONURIA	PAH	Pro281Leu
PHENYLKETONURIA	PAH	Tyr204Cys
PHENYLKETONURIA	PAH	Arg243Gln
PHENYLKETONURIA	PAH	Trp326Ter
PHENYLKETONURIA	PAH	Arg413Pro
PHENYLKETONURIA	PAH	Tyr356Ter
PHENYLKETONURIA	PAH	Gly272Ter
PHENYLKETONURIA	PAH	Ser273Phe
PHENYLKETONURIA	PAH	Leu255Ser
PHENYLKETONURIA	PAH	Ala259Val
PHENYLKETONURIA	PAH	Tyr277Asp
PHENYLKETONURIA	PAH	Phe39Leu
PHENYLKETONURIA	PAH	Ser349Arg
PHENYLKETONURIA	PAH	Leu48Ser
PHENYLKETONURIA	PAH	Glu221Gly
PHENYLKETONURIA	PAH	Arg261Ter
PHENYLKETONURIA	PAH	Arg408Gln
PHENYLKETONURIA	PAH	Phe299Cys
PHENYLKETONURIA	PAH	Ser349Pro
HYPERPHENYLALANINEMIA, NON-PKU MILD	PAH	Ala322Gly
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Asp415Asn
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Ile306Val
PHENYLKETONURIA	PAH	Val388Met
PHENYLKETONURIA	PAH	Pro244Leu
PHENYLKETONURIA	PAH	Met1Ile
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Leu333Phe
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Glu390Gly
PHENYLKETONURIA	PAH	Ser359Ter
HYPERPHENYLALANINEMIA, NON-PKU MILD	PAH	Leu98Ser
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Thr380Met
PHENYLKETONURIA	PAH	Gly46Ser
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Ala47Val
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Ser87Arg
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Arg176Leu

HYPERPHENYLALANINEMIA, NON-PKU	PAH	Val245Ala
PHENYLKETONURIA	PAH	Pro407Leu
PHENYLKETONURIA	PAH	Ile65Thr
HYPERPHENYLALANINEMIA, NON-PKU	PAH	Glu76Gly
Cystic Fibrosis	CFTR	Ser18Arg
Cystic Fibrosis	CFTR	Arg31Cys
Cystic Fibrosis	CFTR	Gln39Ter
Cystic Fibrosis	CFTR	Ala46Asp
Cystic Fibrosis	CFTR	Glu56Lys
Cystic Fibrosis	CFTR	Arg59Lys
Cystic Fibrosis	CFTR	Glu60Ter
Cystic Fibrosis	CFTR	Pro67Leu
Cystic Fibrosis	CFTR	Arg74Trp
Cystic Fibrosis	CFTR	Arg75Ter
Cystic Fibrosis	CFTR	Arg75Gln
Cystic Fibrosis	CFTR	Gly85Glu
Cystic Fibrosis	CFTR	Leu88Ile
Cystic Fibrosis	CFTR	Glu92Lys
Cystic Fibrosis	CFTR	Glu92Ter
Cystic Fibrosis	CFTR	Gln98Ter
Cystic Fibrosis	CFTR	Ile105Ser
Cystic Fibrosis	CFTR	Tyr109Gly
Cystic Fibrosis	CFTR	Asp110His
Cystic Fibrosis	CFTR	Arg117Cys
Cystic Fibrosis	CFTR	Arg117His
Cystic Fibrosis	CFTR	Tyr122Ter
Cystic Fibrosis	CFTR	Ile148Leu
Cystic Fibrosis	CFTR	Ile148Thr
Cystic Fibrosis	CFTR	Gly178Glu
Cystic Fibrosis	CFTR	Leu183Phe
Cystic Fibrosis	CFTR	His199Tyr
Cystic Fibrosis	CFTR	Pro205Ser
Cystic Fibrosis	CFTR	Leu206Trp
Cystic Fibrosis	CFTR	Gln220Ter
Cystic Fibrosis	CFTR	Leu227Arg
Cystic Fibrosis	CFTR	Gly241Glu
Cystic Fibrosis	CFTR	Cys276Ter
Cystic Fibrosis	CFTR	Phe316Leu
Cystic Fibrosis	CFTR	Gly330Ter
Cystic Fibrosis	CFTR	Arg334Trp
Cystic Fibrosis	CFTR	Ile336Lys
Cystic Fibrosis	CFTR	Thr338Ile
Cystic Fibrosis	CFTR	Ser341Pro
Cystic Fibrosis	CFTR	Phe342His
Cystic Fibrosis	CFTR	Arg347His
Cystic Fibrosis	CFTR	Arg347Pro
Cystic Fibrosis	CFTR	Arg352Gln
Cystic Fibrosis	CFTR	Gln359Lys
Cystic Fibrosis	CFTR	Trp361Gly
Cystic Fibrosis	CFTR	Gln378Ala
Cystic Fibrosis	CFTR	Asn386Ile
Cystic Fibrosis	CFTR	Trp401Ter
Cystic Fibrosis	CFTR	Gln414Ter
Cystic Fibrosis	CFTR	Ile444Arg
Cystic Fibrosis	CFTR	Lys447Arg
Cystic Fibrosis	CFTR	Ala455Glu
Cystic Fibrosis	CFTR	Ser466Ter
Cystic Fibrosis	CFTR	Leu467Pro
Cystic Fibrosis	CFTR	Gly473Glu
Cystic Fibrosis	CFTR	Ser489Ter
Cystic Fibrosis	CFTR	Ser492Phe
Cystic Fibrosis	CFTR	Gln493Ter
Cystic Fibrosis	CFTR	Ile507del
Cystic Fibrosis	CFTR	Phe508del
Cystic Fibrosis	CFTR	Tyr515Ter
Cystic Fibrosis	CFTR	Val520Phe
Cystic Fibrosis	CFTR	Gln525Ter
Cystic Fibrosis	CFTR	Gly542Ter
Cystic Fibrosis	CFTR	Ser549Arg
Cystic Fibrosis	CFTR	pSer549Asn
Cystic Fibrosis	CFTR	Gly551Val
Cystic Fibrosis	CFTR	Gly551Ser
Cystic Fibrosis	CFTR	Gly551Asp
Cystic Fibrosis	CFTR	Gln552Ter

Cystic Fibrosis	CFTR	Arg553Ter
Cystic Fibrosis	CFTR	Leu558Ser
Cystic Fibrosis	CFTR	Ala559Thr
Cystic Fibrosis	CFTR	Arg560Lys
Cystic Fibrosis	CFTR	Arg560Thr
Cystic Fibrosis	CFTR	Ala561Glu
Cystic Fibrosis	CFTR	Asp565Met
Cystic Fibrosis	CFTR	Tyr569Asp
Cystic Fibrosis	CFTR	Gly576Ala
Cystic Fibrosis	CFTR	Asp579Gly
Cystic Fibrosis	CFTR	Glu585Ter
Cystic Fibrosis	CFTR	Asp614Gly
Cystic Fibrosis	CFTR	Ser641Arg
Cystic Fibrosis	CFTR	Arg658Lys
Cystic Fibrosis	CFTR	Thr663Arg
Cystic Fibrosis	CFTR	Arg668Cys
Cystic Fibrosis	CFTR	Leu671Ter
Cystic Fibrosis	CFTR	Lys684Ser
Cystic Fibrosis	CFTR	Gln685Thr
Cystic Fibrosis	CFTR	Lys684Asn
Cystic Fibrosis	CFTR	Arg709Ter
Cystic Fibrosis	CFTR	Lys710Ter
Cystic Fibrosis	CFTR	Glu726Arg
Cystic Fibrosis	CFTR	Leu732Ter
Cystic Fibrosis	CFTR	Val739Tyr
Cystic Fibrosis	CFTR	Val754Met
Cystic Fibrosis	CFTR	Trp846Ter
Cystic Fibrosis	CFTR	Tyr849Ter
Cystic Fibrosis	CFTR	Arg851Ter
Cystic Fibrosis	CFTR	Phe861Leu
Cystic Fibrosis	CFTR	Gln890Ter
Cystic Fibrosis	CFTR	Ser912Ter
Cystic Fibrosis	CFTR	Val922Glu
Cystic Fibrosis	CFTR	Leu927Pro
Cystic Fibrosis	CFTR	Ser945Leu
Cystic Fibrosis	CFTR	Ala959His
Cystic Fibrosis	CFTR	Gly970Arg
Cystic Fibrosis	CFTR	Ser977Phe
Cystic Fibrosis	CFTR	Leu997Phe
Cystic Fibrosis	CFTR	Val1001Asp
Cystic Fibrosis	CFTR	Ile1027Thr
Cystic Fibrosis	CFTR	Phe1052Val
Cystic Fibrosis	CFTR	His1054Asp
Cystic Fibrosis	CFTR	Gly1061Arg
Cystic Fibrosis	CFTR	Leu1065Pro
Cystic Fibrosis	CFTR	Arg1066Cys
Cystic Fibrosis	CFTR	Arg1066His
Cystic Fibrosis	CFTR	Gly1069Arg
Cystic Fibrosis	CFTR	Arg1070Trp
Cystic Fibrosis	CFTR	Arg1070Gln
Cystic Fibrosis	CFTR	Phe1074Leu
Cystic Fibrosis	CFTR	Leu1077Pro
Cystic Fibrosis	CFTR	Trp1089Ter
Cystic Fibrosis	CFTR	Tyr1092Ter
Cystic Fibrosis	CFTR	Met1101Lys
Cystic Fibrosis	CFTR	Glu1104Ter
Cystic Fibrosis	CFTR	Asp1152His
Cystic Fibrosis	CFTR	Arg1158Ter
Cystic Fibrosis	CFTR	Arg1162Ter
Cystic Fibrosis	CFTR	Arg1162Leu
Cystic Fibrosis	CFTR	Lys1177Ser
Cystic Fibrosis	CFTR	Thr1179Ile
Cystic Fibrosis	CFTR	Ser1196Ter
Cystic Fibrosis	CFTR	Asp1202Ala
Cystic Fibrosis	CFTR	Trp1204Ter
Cystic Fibrosis	CFTR	Thr1220Lys
Cystic Fibrosis	CFTR	Ser1231Pro
Cystic Fibrosis	CFTR	Ile1234Val
Cystic Fibrosis	CFTR	Ser1235Arg
Cystic Fibrosis	CFTR	Gly1244Glu
Cystic Fibrosis	CFTR	Lys1250Arg
Cystic Fibrosis	CFTR	Ser1251Asn
Cystic Fibrosis	CFTR	Ser1255Pro
Cystic Fibrosis	CFTR	Ser1255Ter

Cystic Fibrosis	CFTR	Leu1258Phe
Cystic Fibrosis	CFTR	Asp1270Asn
Cystic Fibrosis	CFTR	Trp1282Ter
Cystic Fibrosis	CFTR	Ile1295Phe
Cystic Fibrosis	CFTR	Ser1297Phe
Cystic Fibrosis	CFTR	Asn1303Lys
Cystic Fibrosis	CFTR	Gln1313Ter
Cystic Fibrosis	CFTR	Gly1349Asp
Cystic Fibrosis	CFTR	Glu1371Ter
Cystic Fibrosis	CFTR	Cys1400Ter
Cystic Fibrosis	CFTR	Gln1412Ter
Cystic Fibrosis	CFTR	Glu1418Arg
Cystic Fibrosis	CFTR	Ser1435Gly
GALACTOSEMIA	GALT	Met142Lys
GALACTOSEMIA	GALT	Arg333Trp
GALACTOSEMIA	GALT	Val44Met
GALACTOSEMIA	GALT	Gln188Arg
GALACTOSEMIA	GALT	Leu74Pro
GALACTOSEMIA	GALT	Phe171Ser
GALACTOSEMIA	GALT	His319Gln
GALACTOSEMIA	GALT	Ser135Leu
GALACTOSEMIA	GALT	Pro183Thr
GALACTOSEMIA	GALT	Lys285Asn
GALACTOSEMIA	GALT	Glu203Lys
GALACTOSEMIA	GALT	Arg333Gly
GALACTOSEMIA	GALT	Phe194Leu
HOLT-ORAM SYNDROME	TBX5	Glu69Ter
HOLT-ORAM SYNDROME	TBX5	Arg237Gln
HOLT-ORAM SYNDROME	TBX5	Gly80Arg
HOLT-ORAM SYNDROME	TBX5	Arg237Trp
HOLT-ORAM SYNDROME	TBX5	Gln49Lys
HOLT-ORAM SYNDROME	TBX5	Ile54Thr
HOLT-ORAM SYNDROME	TBX5	Tyr136Ter
HYPOCHONDROPLASIA	FGFR3	Asn540Lys
HYPOCHONDROPLASIA	FGFR3	Asn540Thr
HYPOCHONDROPLASIA	FGFR3	Ile538Val
HYPOCHONDROPLASIA	FGFR3	Lys650Asn
HYPOCHONDROPLASIA	FGFR3	Lys650Asn
HYPOCHONDROPLASIA	FGFR3	Lys650Gln
HYPOCHONDROPLASIA	FGFR3	Asn540Ser
HYPOCHONDROPLASIA	FGFR3	Tyr278Cys
HYPOCHONDROPLASIA	FGFR3	Ser84Leu
HYPOCHONDROPLASIA	FGFR3	Gly342Cys
HYPOPHOSPHATASIA, INFANTILE	ALPL	Ala162Thr
HYPOPHOSPHATASIA, INFANTILE	ALPL	Arg54Cys
HYPOPHOSPHATASIA, INFANTILE	ALPL	Asp277Ala
HYPOPHOSPHATASIA, INFANTILE	ALPL	Arg54Pro
HYPOPHOSPHATASIA, INFANTILE	ALPL	Gln190Pro
HYPOPHOSPHATASIA, INFANTILE	ALPL	Ala16Val
HYPOPHOSPHATASIA, INFANTILE	ALPL	Tyr419His
HYPOPHOSPHATASIA, INFANTILE	ALPL	Glu174Lys
HYPOPHOSPHATASIA, INFANTILE	ALPL	Asp378Val
HYPOPHOSPHATASIA, INFANTILE	ALPL	Gly317Asp
HYPOPHOSPHATASIA, INFANTILE	ALPL	Phe310Leu
HYPOPHOSPHATASIA, CHILDHOOD	ALPL	Arg119His
HYPOPHOSPHATASIA, CHILDHOOD	ALPL	Gly145Val
HYPOPHOSPHATASIA, CHILDHOOD	ALPL	Ala99Thr
HYPOPHOSPHATASIA, INFANTILE	ALPL	Asn400Ser
HYPOPHOSPHATASIA, INFANTILE	ALPL	Gly439Arg
HYPOPHOSPHATASIA, INFANTILE	ALPL	Glu281Lys
HYPOPHOSPHATASIA, CHILDHOOD	ALPL	Gly232Val
HYPOPHOSPHATASIA, INFANTILE	ALPL	Ala176Thr
HYPOPHOSPHATASIA, INFANTILE	ALPL	Arg272Cys
LISSENCEPHALY, X-LINKED, 1	DCX	Asp62Asn
LISSENCEPHALY, X-LINKED, 1	DCX	Arg192Trp
LISSENCEPHALY, X-LINKED, 1	DCX	Tyr125His
LISSENCEPHALY, X-LINKED, 1	DCX	Arg59Leu
LISSENCEPHALY, X-LINKED, 1	DCX	Thr203Arg
LISSENCEPHALY, X-LINKED, 1	DCX	Ser47Arg
LISSENCEPHALY, X-LINKED, 1	DCX	Arg196His
LISSENCEPHALY, X-LINKED, 1	DCX	Ala71Ser
LISSENCEPHALY 3	TUBA1A	Arg264Cys
LISSENCEPHALY 3	TUBA1A	Arg402His
LISSENCEPHALY 3	TUBA1A	Ile188Leu
LISSENCEPHALY 3	TUBA1A	Pro263Thr
LISSENCEPHALY 3	TUBA1A	Ser419Leu
LISSENCEPHALY 3	TUBA1A	Leu397Pro

LISSENCEPHALY 3	TUBA1A	Arg422Cys
LISSENCEPHALY 3	TUBA1A	Arg422His
LISSENCEPHALY 3	TUBA1A	Ile5Leu
JOUBERT SYNDROME 3	AHI1	Arg351Ter
JOUBERT SYNDROME 3	AHI1	Arg435Ter
JOUBERT SYNDROME 3	AHI1	Val443Asp
JOUBERT SYNDROME 3	AHI1	Arg589Ter
JOUBERT SYNDROME 3	AHI1	Arg723Gln
JOUBERT SYNDROME 3	AHI1	Arg329Ter
JOUBERT SYNDROME 3	AHI1	Arg495His
JOUBERT SYNDROME 3	AHI1	Arg351Leu
JOUBERT SYNDROME 5	CEP290	Gly1890Ter
JOUBERT SYNDROME 5	CEP290	Trp7Cys
JOUBERT SYNDROME 5	CEP290	Lys1575Ter
JOUBERT SYNDROME 6	TMEM67	Tyr513Cys
JOUBERT SYNDROME 6	TMEM67	Gly545Glu
JOUBERT SYNDROME 6	TMEM67	Gln44Ter
JOUBERT SYNDROME 6	TMEM67	Met252Thr
JOUBERT SYNDROME 6	TMEM67	Pro358Leu
JOUBERT sindrom.8	ALR13B	Trp82Ter
JOUBERT sindrom.8	ALR13B	Arg200Cys
JOUBERT sindrom.8	ALR13B	Lys233Ter
JOUBERT SYNDROME 7	RPGRIP1L	Lys233Ter
JOUBERT SYNDROME 7	RPGRIP1L	Thr615Pro
JOUBERT SYNDROME 7	RPGRIP1L	Gln253Ter
JOUBERT SYNDROME 7	RPGRIP1L	Ala695Pro
JOUBERT SYNDROME 7	RPGRIP1L	Gln684Ter
JOUBERT SYNDROME 9	CC2D2A	Pro1122Ser
JOUBERT SYNDROME 9	CC2D2A	Arg1528Cys
JOUBERT SYNDROME 9	CC2D2A	Arg950Ter
JOUBERT SYNDROME 9/15, DIGENIC	CC2D2A	Glu1447Ala
JOUBERT SYNDROME 2	TMEM216	Arg73Leu
JOUBERT SYNDROME 2	TMEM216	Arg73His
KABUKI	KMT2D	Arg5179His
KABUKI	KMT2D	Lys4527Ter
KABUKI	KMT2D	Arg5454Ter
KABUKI	KMT2D	Thr5464Met
MARFAN	FBN1	Arg1137Pro
MARFAN	FBN1	Cys2307Ser
MARFAN	FBN1	Trp2756Ter
MARFAN	FBN1	Cys1249Ser
MARFAN	FBN1	Cys1663Arg
MARFAN	FBN1	Cys2221Ser
MARFAN	FBN1	Tyr2113Ter
MARFAN	FBN1	Asn2144Ser
MARFAN	FBN1	Asn548Ile
MARFAN	FBN1	Asp723Ala
MARFAN	FBN1	Cys1074Arg
MARFAN	FBN1	Arg2776Ter
MARFAN	FBN1	Arg122Cys
MARFAN	FBN1	Gly1127Ser
MARFAN	FBN1	Cys1223Tyr
MARFAN	FBN1	Cys1117Tyr
MARFAN	FBN1	Cys1242Tyr
MARFAN	FBN1	Lys1043Arg
MARFAN	FBN1	Asn1131Tyr
MARFAN	FBN1	Cys1265Arg
MARFAN	FBN1	Arg1170His
MARFAN	FBN1	Arg529Ter
MARFAN	FBN1	Gly985Glu
MARFAN	FBN1	Gly1013Arg
MARFAN	FBN1	Glu1073Lys
MARFAN	FBN1	Tyr754Cys
MARFAN	FBN1	Arg240Cys
MARFAN	FBN1	Cys1032Tyr
MARFAN	FBN1	Cys1129Tyr
MARFAN	FBN1	Cys1221Tyr
MARFAN	FBN1	Cys1086Tyr
MARFAN	FBN1	Arg485Cys
MARFAN	FBN1	Arg2576Cys
MECKEL	MKS1	Arg158Ter
MECKEL	MKS1	Arg166Trp
MECKEL	MKS1	Glu277Ter
MECKEL	MKS1	Gln350Ter
MICROCEFALIA	ASPM	Gln3060Ter
MICROCEFALIA	ASPM	Trp1326Ter

MICROCEFALIA	ASPM	Arg117Ter
MICROCEFALIA	ASPM	Tyr2063Ter
MICROCEFALIA	ASPM	Arg797Ter
MUCOLIPIDOSI	MCOLN1	Arg321Ter
MUCOLIPIDOSI	MCOLN1	Asp362Tyr
MUCOLIPIDOSI	MCOLN1	Arg102Ter
MUCOLIPIDOSI	MCOLN1	Arg403Cys
NAIL-PATELLA	LMX1B	Arg198Ter
NAIL-PATELLA	LMX1B	Cys95Phe
NAIL-PATELLA	LMX1B	Gln59Ter
NAIL-PATELLA	LMX1B	Arg208Ter
NAIL-PATELLA	LMX1B	Arg200Gln
NAIL-PATELLA	LMX1B	Arg226Ter
NOONAN	PTPN11	Asn308Asp
NOONAN	PTPN11	Asn308Ser
NOONAN	PTPN11	Tyr279Cys
NOONAN	PTPN11	Thr468Met
NOONAN	PTPN11	Ser502Thr
NOONAN	PTPN11	Phe285Ser
NOONAN	PTPN11	Gln79Arg
NOONAN	PTPN11	Thr411Met
NOONAN	PTPN11	Ala461Thr
NOONAN	PTPN11	Gly464Ala
NOONAN	PTPN11	Gln510Pro
NOONAN	PTPN11	Gln510Arg
NOONAN	PTPN11	Arg138Ter
NOONAN	PTPN11	Thr2Ile
NOONAN	SOS1	Thr266Lys
NOONAN	SOS1	Met269Arg
NOONAN	SOS1	Arg552Gly
NOONAN	SOS1	Arg552Ser
NOONAN	SOS1	Trp432Arg
NOONAN	KRAS	Thr58Ile
NOONAN	KRAS	Val14Ile
NOONAN	KRAS	Val152Gly
NOONAN	KRAS	Asp153Val
NOONAN	KRAS	Lys5Glu
NOONAN	KRAS	Gly60Ser
NOONAN	RAF1	Ser257Leu
NOONAN	RAF1	Pro261Ser
NOONAN	BRAF	Thr241Met
NOONAN	BRAF	Thr241Arg
NOONAN	BRAF	Trp531Cys
NOONAN	NRAS	Gly60Glu
NOONAN	CBL	Gln367Pro
NOONAN	CBL	Lys382Glu
NOONAN	CBL	Asp390Tyr
NOONAN	CBL	Arg420Gln
NOONAN	CBL	Tyr371His
NOONAN	CBL	Cys384Arg
NOONAN	CBL	Cys396Arg
NOONAN	CBL	Tyr371Cys
NOONAN	SHH	Gly31Arg
OLOPROSENCEFALIA	SHH	Gln100Ter
OLOPROSENCEFALIA	SHH	Lys105Ter
OLOPROSENCEFALIA	SHH	Trp117Gly
OLOPROSENCEFALIA	SHH	Trp117Arg
OLOPROSENCEFALIA	SHH	Val224Glu
OLOPROSENCEFALIA	SHH	Ala226Thr
OLOPROSENCEFALIA	SHH	Glu284Ter
OLOPROSENCEFALIA	SHH	Ala384Thr
OLOPROSENCEFALIA	SHH	Gly290Asp
OLOPROSENCEFALIA	SHH	Pro424Ala
OLOPROSENCEFALIA	SHH	Asp88Val
OLOPROSENCEFALIA	SHH	Glu256Ter
OLOPROSENCEFALIA	SHH	Trp128Ter
OLOPROSENCEFALIA	SHH	Asn115Lys
OLOPROSENCEFALIA	SIX3	Leu226Val
OLOPROSENCEFALIA	SIX3	Gly69Asp
OLOPROSENCEFALIA	SIX3	Trp113Cys
OLOPROSENCEFALIA	SIX3	Glu129Ter
OLOPROSENCEFALIA	SIX3	Gly37Cys
OSTEOGENESI IMPERFETTA	COL1A1	Gly85Arg
OSTEOGENESI IMPERFETTA	COL1A1	Gly1017Cys

OSTEOGENESI IMPERFETTA, TIPO I	COL1A1	Gly94Cys
OSTEOGENESI IMPERFETTA, TIPO I	COL1A1	Gly178Cys
OSTEOGENESI IMPERFETTA, TIPO I	COL1A1	Arg963Ter
OSTEOGENESI IMPERFETTA, TIPO I	COL1A1	Gly901Ser
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly391Arg
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly97Asp
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly541Asp
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly559Asp
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly673Asp
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly667Arg
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly691Cys
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly718Cys
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly847Arg
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly883Asp
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly904Cys
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly913Ser
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly988Cys
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly1009Ser
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly1003Ser
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly637Val
OSTEOGENESI IMPERFETTA, TIPO II	COL1A1	Gly565Val
OSTEOGENESI IMPERFETTA, TIPO II	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
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Gly415Ser
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly976Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly805Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly259Cys
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly472Cys
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly421Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Gly586Val
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly694Arg
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly580Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Ala1387Val
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly865Ser
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly907Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly502Ser
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A2	Gly547Asp
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Trp94Cys
OSTEOGENESIS IMPERFECTA, TYPE II	COL1A1	Gly748Cys
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly862Ser
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly661Ser
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly526Cys
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly154Arg
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly844Ser
OSTEOGENESIS IMPERFECTA, TYPE III/IV	COL1A1	Gly415Cys
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly352Ser
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly1006Ala
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly586Val
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly751Ser
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly277Tyr
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A1	Gly76Glu
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly859Ser
OSTEOGENESIS IMPERFECTA, TYPE III	COL1A2	Gly1090Asp
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A1	Gly832Ser
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A1	Gly175Cys
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A2	Gly646Cys
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A2	Gly586Val
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A2	Gly1012Arg
OSTEOGENESIS IMPERFECTA, TYPE IV	COL1A2	Gly379Ala
OSTEOGENESIS IMPERFECTA, TYPE VII	CRTAP	Gln276Ter
OSTEOGENESIS IMPERFECTA, TYPE VII	CRTAP	Tyr187Ter
OSTEOGENESI IMPERFETTA	LEPRE1	Tyr552Ter
OSTEOGENESI IMPERFETTA	LEPRE1	Arg368Ter
POLIMICROGIRIA	TUBB2B	Ser172Pro
POLIMICROGIRIA	TUBB2B	Leu228Pro
POLIMICROGIRIA	TUBB2B	Phe265Leu
POLIMICROGIRIA	TUBB2B	Asp417Asn
POLIMICROGIRIA	TUBB2B	Asn256Ser
POLIMICROGIRIA	TUBB2B	Leu117Pro
RENE POLICISTICO dell'ADULTO	PKD1	Gln1273Ter
RENE POLICISTICO dell'ADULTO	PKD1	Arg4227Ter

RENE POLICISTICO dell'ADULTO	PKD1	Gln3837Ter
RENE POLICISTICO dell'ADULTO	PKD1	Cys4086Ter
RENE POLICISTICO dell'ADULTO	PKD1	Tyr3818Ter
RENE POLICISTICO dell'ADULTO	PKD1	Arg324Leu
RENE POLICISTICO dell'ADULTO	PKD1	Leu845Ser
RENE POLICISTICO dell'ADULTO	PKD1	Gln1922Ter
RENE POLICISTICO dell'ADULTO	PKD1	Trp4139Ter
RENE POLICISTICO dell'ADULTO	PKD2	Trp380Ter
RENE POLICISTICO dell'ADULTO	PKD2	Arg742Ter
RENE POLICISTICO dell'ADULTO	PKD2	Gln405Ter
RENE POLICISTICO dell'ADULTO	PKD2	Arg464Ter
RENE POLICISTICO dell'ADULTO	PKD2	Asp511Val
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Thr36Met
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Ser1664Phe
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Ser3018Phe
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Arg2671Ter
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Ile3553Thr
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Arg496Ter
RENE POLICISTICO AUTOSOMICO RECESSIVO	PKHD1	Val3471Gly
PFEIFFER	FGFR1	Cys342Tyr
PFEIFFER	FGFR1	Cys342Arg
PFEIFFER	FGFR1	Thr341Pro
PFEIFFER	FGFR1	Trp290Cys
PFEIFFER	FGFR1	Ser351Cys
PFEIFFER	FGFR1	Ser267Pro
PFEIFFER	FGFR1	Trp290Cys
PFEIFFER	FGFR1	Glu565Ala
PFEIFFER	FGFR1	Asp321Ala
RETT	MECP2	Arg133Cys
RETT	MECP2	Phe155Ser
RETT	MECP2	Arg270Ter
RETT	MECP2	Thr158Met
RETT	MECP2	Arg106Trp
RETT	MECP2	Arg294Ter
RETT	MECP2	Arg306Cys
RETT	MECP2	Arg168Ter
RETT	MECP2	Arg255Ter
RETT	MECP2	Glu455Ter
RETT	MECP2	Leu100Val
RETT	MECP2	Pro152Ala
SAETHRE-CHOTZEN	TWIST1	Tyr103Ter
SAETHRE-CHOTZEN	TWIST1	Gln119Pro
SAETHRE-CHOTZEN	TWIST1	Tyr107Ter
SAETHRE-CHOTZEN	TWIST1	Ser127Ter
SAETHRE-CHOTZEN	TWIST1	Glu130Ter
SAETHRE-CHOTZEN	TWIST1	Leu135Pro
SAETHRE-CHOTZEN	TWIST1	Glu181Ter
SAETHRE-CHOTZEN	TWIST1	Ile156Val
SECKEL	ATR	Asp1879Tyr
SECKEL	ATR	Met1159Ile
SMITH-LEMLI-OPITZ	DHCR7	His119Leu
SMITH-LEMLI-OPITZ	DHCR7	Gly244Arg
SMITH-LEMLI-OPITZ	DHCR7	Trp248Cys
SMITH-LEMLI-OPITZ	DHCR7	Thr93Met
SMITH-LEMLI-OPITZ	DHCR7	Trp151Ter
SMITH-LEMLI-OPITZ	DHCR7	Val326Leu
SMITH-LEMLI-OPITZ	DHCR7	Trp37Ter
SMITH-LEMLI-OPITZ	DHCR7	Arg352Trp
SMITH-LEMLI-OPITZ	DHCR7	Arg404Cys
SMITH-LEMLI-OPITZ	DHCR7	Thr289Ile
SMITH-LEMLI-OPITZ	DHCR7	Tyr280Cys
SMITH-LEMLI-OPITZ	DHCR7	Met1Leu
SMITH-LEMLI-OPITZ	DHCR7	Glu448Lys
SMITH-LEMLI-OPITZ	DHCR7	Phe284Leu
SMITH-LEMLI-OPITZ	DHCR7	Met1Val
SMITH-LEMLI-OPITZ	DHCR7	Arg352Gln
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Met34Thr
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Trp24Ter
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Trp77Arg
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Glu47Ter
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Glu118del
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Arg184Pro
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Arg143Trp
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Leu90Pro

SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Val37Ile
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Asp159Val
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Val84Leu
SORDITA' CONGENITA AUT. RECESSIVA	GJB2	Val84Met
SOTOS	NSD1	Ser437Ter
SOTOS	NSD1	His2143Glu
SOTOS	NSD1	Cys2183Ser
SOTOS	NSD1	Arg1320Ter
SOTOS	NSD1	Cys2202Tyr
TAY-SACHS DISEASE	HEXA	Glu482Lys
TAY-SACHS DISEASE	HEXA	Arg178Cys
TAY-SACHS DISEASE	HEXA	Arg170Gln
TAY-SACHS DISEASE	HEXA	Trp420Cys
TAY-SACHS DISEASE	HEXA	Gly250Asp
TAY-SACHS DISEASE	HEXA	Ser210Phe
TAY-SACHS DISEASE	HEXA	Arg137Ter
TAY-SACHS DISEASE	HEXA	Arg393Ter
TAY-SACHS DISEASE	HEXA	Trp26Ter
TAY-SACHS DISEASE	HEXA	Arg178Leu
TAY-SACHS DISEASE	HEXA	Met1Val
TAY-SACHS DISEASE	HEXA	Trp485Arg
TAY-SACHS DISEASE	HEXA	Tyr180Ter
TAY-SACHS DISEASE	HEXA	Asp258His
TAY-SACHS DISEASE	HEXA	Arg170Trp
TAY-SACHS DISEASE	HEXA	Phe211Ser
TAY-SACHS DISEASE	HEXA	Leu127Arg
TAY-SACHS DISEASE	HEXA	His204Arg
TAY-SACHS DISEASE	HEXA	Met301Arg
TAY-SACHS DISEASE	HEXA	Gly454Ser
TAY-SACHS DISEASE	HEXA	Leu39Arg
TAY-SACHS DISEASE	HEXA	Trp392Ter
TAY-SACHS DISEASE	HEXA	Leu451Val
TIROSINEMIA	FAH	Asn16Ile
TIROSINEMIA	FAH	Ala134Asp
TIROSINEMIA	FAH	Glu357Ter
TIROSINEMIA	FAH	Glu364Ter
TIROSINEMIA	FAH	Arg341Trp
TIROSINEMIA	FAH	Glu337Ser
TIROSINEMIA	FAH	Arg381Gly
TIROSINEMIA	FAH	Trp262Ter
TIROSINEMIA	FAH	Gln279Arg
TREACHER COLLINS	TCOF1	Gln252Ter
TREACHER COLLINS	TCOF1	Tyr50Cys
TREACHER COLLINS	TCOF1	Arg911Ter
WILSON	ATP7B	His714Gln
WILSON	ATP7B	Asn915Ser
WILSON	ATP7B	His1069Gln
WILSON	ATP7B	Gly1267Arg
WILSON	ATP7B	Arg778Leu
WILSON	ATP7B	Asp765Asn
WILSON	ATP7B	Gly943Ser
WILSON	ATP7B	Arg919Gly
WILSON	ATP7B	Ala874Val
WILSON	ATP7B	Asn1270Ser
WILSON	ATP7B	Arg969Gln
WILSON	ATP7B	Thr766Arg
WILSON	ATP7B	Met645Arg
WILSON	ATP7B	Gly1176Arg
WILSON	ATP7B	Ile1148Thr
WILSON	ATP7B	Gln289Ter
WILSON	ATP7B	Leu708Pro
WILSON	ATP7B	Gly691Arg
WILSON	ATP7B	Ile1148Thr
ZELLWEGER	PEX1	Leu664Pro