LBM/UMAI/0284 V1

Diagnostic DES FENTES LABIO-PALATINES, CHROMATINOPATHIES +/- DEFICIENCE INTELLECTUELLE

GENETIQUE						PHENOTYPE				
Gene	**HGNC ID**	**Nom**	**OMIM**	**Locus**	**Transcrit RefSeq**	**Hérédité**	**Disease**	**Nom**	**OMIM**	**Aliases**
ABCC9	HGNC:60		601439	12p12.1	NM_020297.3	Dominante		CANTU SYNDROME	239850	
ANKRD11	HGNC:21316		611192	16q24.3	NM_001256182.1	Dominante	KBGS	KBG SYNDROME	148050	
ARID1A	HGNC:11110		603024	1p36.11	NM_006015.4	Dominante	MRD14	MENTAL RETARDATION 14	614607	
ARID1B	HGNC:18040		614556	6q25.3	NM_020732.3	Dominante	MRD12	MENTAL RETARDATION 12	614562	
ATP6V1B2	HGNC:854		606939	8p21.3	NM_001693.3	Dominante	DDODS	DEAFNESS, CONGENITAL, WITH ONYCHODYSTROPHY,	124480	
BRPF1	HGNC:14255		602410	3p25.3	NM_001003694.1	Recessive	CFDS	CEREBELLOFACIODENTAL SYNDROME	616202	
CHD7	HGNC:20626		608892	8q12.2	NM_017780.3	Dominante	HHS	CHARGE SYNDROME	214800	
COLEC11	HGNC:17213		612502	2p25.3	NM_024027.4	Recessive	3MC2	3MC SYNDROME 2	265050	
CREBBP	HGNC:2348		600140	16p13.3	NM_004380.2	Dominante	RSTS1	RUBINSTEIN-TAYBI SYNDROME 1	180849	
DVL1	HGNC:3084		601365	1p36.33	NM_004421.2	Dominante	DRS2	ROBINOW SYNDROME 2	616331	
DVL3	HGNC:3087		601368	3q27.1	NM_004423.3	Dominante	DRS3	ROBINOW SYNDROME 3	616894	
EDN1	HGNC:3176		131240	6p24.1	NM_001955.4	Recessive	ARCND3	AURICULOCONDYLAR SYNDROME 3	615706	
EDNRA	HGNC:3179		131243	4q31.22-q31	NM_001957.3	Dominante	MFDA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA	616367	
EFTUD2	HGNC:30858		603892	17q21.31	NM_004247.3	Dominante	MFDGA	MANDIBULOFACIAL DYSOSTOSIS, GUION-ALMEIDA TYPE	610536	
EHMT1	HGNC:24650		607001	9q34.3	NM_024757	Dominante	KLEFS1	KLEEFSTRA SYNDROME	610253	
EIF4A3	HGNC:18683		608546	17q25.3	NM_014740.3	Recessive	RCPS	RICHIERI-COSTA-PEREIRA SYNDROME	268305	
EP300	HGNC:3373		602700	22q13.2	NM_001429.3	Dominante	RSTS2	RUBINSTEIN-TAYBI SYNDROME 2	613684	
FGD1	HGNC:3663		300546	Xp11.22	NM_004463.2	XLR	AAS	AARSKOG-SCOTT SYNDROME	305400	
FLNA	HGNC:3754		300017	Xq28	NM_001110556.1	XLD	OPD1	OTOPALATODIGITALSYNDROME,TYPE I	311300	
FLNA	HGNC:3754		300017	Xq28	NM_001110556.1	XLD	OPD2	OTOPALATODIGITALSYNDROME,TYPE II	304120	
FLNB	HGNC:3755		603381	3p14.3	NM_001164317.1	Dominante	AOI	ATELOSTEOGENESIS,TYPEI	108720	
FLNB	HGNC:3755		603381	3p14.3	NM_001164317.1	Dominante	AOIII	ATELOSTEOGENESIS,TYPEIII	108721	
GNAI3	HGNC:4387		139370	1p13.3	NM_006496.3	Dominante	ARCND1	AURICULOCONDYLAR SYNDROME 1	602483	
GRHL3	HGNC:25839		608317	1p36.11	NM_198174.2	Dominante	VWS2	VAN DER WOUDE SYNDROME 2	606713	
HDAC4	HGNC:14063		605314	2q37.3	NM_006037.3	Gène candidat	BDMR	BRACHYDACTYLY-MENTAL RETARDATION SYNDROME	600430	
HDAC8	HGNC:13315		300269	Xq13.1	NM_018486.2	XLD	CDLS5	CORNELIA DE LANGE SYNDROME 5	300882	
IRF6	HGNC:6121		607199	1q32.2	NM_006147.3	Dominante	VWS1	VAN DER WOUDE SYNDROME 1	119300	

KANSL1	HGNC:24565		612452	17q21.31	NM_001193466.1	Dominante	KDVS	KOOLEN-DE VRIES SYNDROME	610443		
KAT6A	HGNC:13013		601408	8p11.21	NM_001099412	Dominante	MRD32	MENTAL RETARDATION, AUTOSOMAL DOMINANT 32	616268		
KAT6B	HGNC:17582		605880	10q22.2	NM_012330.3	Dominante	SBBYSS	OHDO SYNDROME, SBBYS VARIANT	603736		
KCNH1	HGNC:6250		603305	1q32.2	NM_172362.2	Dominante	TMBTS	TEMPLE-BARAITSER SYNDROME	611816		
KCNJ8	HGNC:6269		600935	12p12.1	NM_004982.3	Recessive	SIDS	SUDDEN INFANT DEATH SYNDROME	272120		
KDM1A	HGNC:29079		609132	1p36.12	NM_001009999.2	Dominante	CPRF	CLEFT PALATE, PSYCHOMOTOR RETARDATION, AND DISTINCTIVE FACIAL FEATURES	616728		
KDM5A	HGNC:9886		180202	12p13.33	NM_001042603.2	Gène candidat					
KDM5B	HGNC:18039		605393	1q32.1	NM_006618.4	Gène candidat					
KDM5C	HGNC:11114		314690	Xp11.22	NM_004187.3	XLR	MRXSCJ	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CLAES-JENSEN TYPE	300534		
KDM6A	HGNC:12637		300128	Xp11.3	NM_021140.3	Dominante	KABUK2	KABUKI SYNDROME 2	300867		
KMT2A	HGNC:7132		159555	11q23.3	NM_001197104.1	Dominante	WDSTS	WIEDEMANN-STEINER SYNDROME	605130		
KMT2D	HGNC:7133		602113	12q13.12	NM_003482.3	Dominante	KABUK1	KABUKI SYNDROME 1	147920		
MASP1	HGNC:6901		600521	3q27.3	NM_139125.3	Recessive	3MC1	3MC SYNDROME	257920		
MED13L	HGNC:22962		608771	12q24.21	NM_015335.4	Dominante	MRFACD	MENTAL RETARDATION AND DISTINCTIVE FACIAL FEATURES WITH OR WITHOUT CARDIAC DEFECTS	616789		
MID1	HGNC:7095		300552	Xp22	NM_000381.3	XLR	GBBB1	OPITZ GBBB SYNDROME, X-LINKED	300000		
NIPBL	HGNC:28862		608667	5p13.2	NM_133433.3	XLR	CDLS1	CORNELIA DE LANGE SYNDROME 1	122470		
PHF21A	HGNC:24156		608325	11p11.2	NM_001101802.1	Gène candidat					
PHF6	HGNC:18145		300414	Xq26.2	NM_032458.2	XLR	BFLS	BORJESON-FORSSMAN-LEHMANN SYNDROME	301900		
PHF8	HGNC:20672		300560	Xp11.22	NM_001184896.1	XLR	MRXSSD	SIDERIUS X-LINKED MENTAL RETARDATION SYNDROME	300263		
PLCB4	HGNC:9059		600810	20p12.3-p12	NM_000933.3	Recessive, Dominante	ARCND2	AURICULOCONDYLAR SYNDROME 2	614669		
POLR1A	HGNC:17264		616404	2p11.2	NM_015425.4	Dominante	AFDCIN	ACROFACIAL DYSOSTOSIS, CINCINNATI TYPE	616462		
POLR1C	HGNC:20194		610060	6p21.1	NM_203290.3	Recessive	TCS3	TREACHER COLLINS SYNDROME 3	248390		
POLR1D	HGNC:20422		613715	13q12.2	NM_015972.3	Recessive, Dominante	TCS2	TREACHER COLLINS SYNDROME 2	613717		
RAD21	HGNC:9811		606462	8q24.11	NM_006265.1	Dominante	CDLS4	CORNELIA DE LANGE SYNDROME 4	614701		
RBM10	HGNC:9896		300080	Xp11.3	NM_005676.4	XLR	TARPS	TARP SYNDROME	311900		
RIPK4	HGNC:496		605706	21q22.3	NM_020639.2	Recessive	BPS	BARTSOCAS-PAPAS SYNDROME	263650		
ROR2	HGNC:10257		602337	9q22.31	NM_004560.3	Recessive	RRS	ROBINOW SYNDROME, AUTOSOMAL RECESSIVE	268310		
SATB2	HGNC:21637		608148	2q33.1	NM_015265.3	Dominante	GLASS	GLASS SYNDROME	612313		
SETD1A	HGNC:29010		611052	16p11.2	NM_014712.2	Gène candidat			611052		
SETD5	HGNC:25566		615743	3p25.3	NM_001080517.2	Dominante	MRD23	MENTAL RETARDATION, AUTOSOMAL DOMINANT 23	615761		
SF3B4	HGNC:10771		605593	1q21.2	NM_005850.4	Dominante	AFD1	ACROFACIAL DYSOSTOSIS 1, NAGER TYPE	154400		

SMARCA2	HGNC:11098		600014	9p24.3	NM_003070.4	Dominante	NCBRS	NICOLAIDES-BARAITSERSYNDROME	601358		
SMARCA4	HGNC:11100		603254	19p13.2	NM_001128849.1	Dominante	MRD16	MENTALRETARDATION, AUTOSOMALDOMINANT16	614609		
SMARCB1	HGNC:11103		601607	22q11.23	NM_003073.4	Dominante	CSS3	COFFIN-SIRIS SYNDROME 3	614608		
SMARCE1	HGNC:11109		603111	17q21.2	NM_003079.4	Dominante	CSS5	COFFIN-SIRIS SYNDROME 5	616938		
SMC1A	HGNC:11111		300040	Xp11.22	NM_006306.3	XLD	CDLS2	CORNELIADELANGESYNDROME2	300590		
SMC3	HGNC:2468		606062	10q25.2	NM_005445.3	Dominante	CDLS3	CORNELIADELANGESYNDROME3	610759		
SNRPB	HGNC:11153		182282	20p13	NM_198216.1	Dominante	CCMS	CEREBROCOSTOMANDIBULAR SYNDROME	117650		
SOX9	HGNC:11204		608160	17q24.3	NM_000346.3	Dominante	CMD1; CMPD1	CAMPOMELIC DYSPLASIA	114290		
SPECC1L	HGNC:29022		614140	22q11.23	NM_015330.4	Dominante	OBLFC1	FACIALCLEFTING,OBLIQUE,1	600251		
SRCAP	HGNC:16974		611421	16p11.2	NM_006662.2	Dominante	FLHS	FLOATING-HARBORSYNDROME	136140		
TBX1	HGNC:11592		602054	22q11.21	NM_080647.1	Dominante	DGS	DIGEORGESYNDROME	188400		
TBX22	HGNC:11600		300307	Xq21.1	NM_016954.2	Gène candidat	CPX	CLEFT PALATE WITH OR WITHOUT ANKYLOGLOSSIA, X-LINKED	303400		
TCOF1	HGNC:11654		606847	5q32-q33.1	NM_001135243.1	Dominante	TCS1	TREACHERCOLLINSSYNDROME1	154500		
TFAP2A	HGNC:11742		107580	6p24.3	NM_003220.2	Dominante	BOFS	BRANCHIOOCULOFACIALSYNDROME	113620		
TGDS	HGNC:20324		616146	13q32.1	NM_014305.3	Recessive	CATMANS	CATEL-MANZKE SYNDROME	616145		
TP63	HGNC:15979		603273	3q28	NM_003722.4	Dominante	EEC3	ECTRODACTYLY, ECTODERMAL DYSPLASIA, AND CLEFT LIP/PALATE SYNDROME 3	604292		
TXNL4A	HGNC:30551		611595	18q23	NM_006701.4	Recessive	BMKS	BURN-MCKEOWN SYNDROME	608572		
WNT5A	HGNC:12784		164975	3p14.3	NM_003392.4	Dominante	DRS1	ROBINOWSYNDROME, AUTOSOMALDOMINANT	180700		
YAP1	HGNC:16262		606608	11q22.1	NM_001130145.2	Dominante	COB1	COLOBOMA, OCULAR, WITH OR WITHOUT HEARING IMPAIRMENT, CLEFT LIP/PALATE, AND/OR MENTAL RETARDATION	120433		