LBM/UMAI/0284 V2

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		
ARID2	HGNC:18037		609539	12q12	NM_152641.4	Dominante	CSS6	COFFIN-SIRIS SYNDROME 6			
ARID1A	HGNC:11110		603024	1p36.11	NM_006015.4	Dominante	CSS2	COFFIN-SIRIS SYNDROME 2	614607		
AMER1	HGNC:26837		300647	Xq11.2	NM_152424.4	XLD	OSCS	OSTEOPATHIA STRIATA WITH CRANIAL SCLEROSIS	300373		
ARID1B	HGNC:18040		614556	6q25.3	NM_020732.3	Dominante	CSS1	COFFIN-SIRIS SYNDROME 1	614562		
ATP6V1B2	HGNC:854		606939	8p21.3	NM_001693.3	Dominante	DDODS	DEAFNESS, CONGENITAL, WITH ONYCHODYSTROPHY,	124480		
BMP2	HGNC:1069		112261	20p12.3	NM_001200.4	Dominante	SSFSC	SHORT STATURE, FACIAL DYSMORPHISM, AND SKELETAL ANOMALIES WITH OR WITHOUT CARDIAC ANOMALIES	617877		
BRPF1	HGNC:14255		602410	3p25.3	NM_001003694.1	Recessive	CFDS	CEREBELLOFACIODENTAL SYNDROME	616202		
CDK13	HGNC:1733		603309	7p14.1	NM_003718.4	Dominante	CHDFIDD	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	617360		
CHD7	HGNC:20626		608892	8q12.2	NM_017780.3	Dominante	HHS	CHARGE SYNDROME	214800		
COL2A1	HGNC:2200		120140	12q13.11	NM_001844.5	Dominante	STL1	Stickler syndrome 1	108300		
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		
DPF2	HGNC:9964		601671	11q13.1	NM_006268.4	Dominante	CSS7	COFFIN-SIRIS SYNDROME 7	618027		
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-q31.23	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		
FOXC2	HGNC:3801		602402	16q24.1	NM_005251.3	Dominante	LPHDST	LYMPHEDEMA-DISTICHIASIS SYNDROME	602402		
FOXE1	HGNC:3806		602617	9q22.33	NM_004473.4	Dominante		HYPOTHYROIDISM, THYROIDAL OR ATHYROIDAL, WITH SPIKY HAIR AND CLEFT PALATE	241850		
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		

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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		
HOXA2	HGNC:5103		604685	7p15.2	NM_006735.4	Recessive, Dominante		MICROTIA, HEARING IMPAIRMENT, AND CLEFT PALATE	612290		
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		
KCNJ2	HGNC:6263		600681	17q24.3	NM_000891.3	Dominante	ATS	ANDERSEN-TAWIL SYNDROME	170390		
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	Recessive	MRT65	MENTAL RETARDATION, AUTOSOMAL RECESSIVE 65	618109		
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		
MSX1	HGNC:7391		142983	4p16.2	NM_002448.3	Dominante	OFC5/ STHAG1	OROFACIAL CLEFT 5/ TOOTH AGENESIS, SELECTIVE, 1	608874/106600		
NECTIN1	HGNC:9706		600644	11q23.3	NM_002855.5	Recessive	CLPED1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME	225060		
NIPBL	HGNC:28862		608667	5p13.2	NM_133433.3	XLR	CDLS1	CORNELIA DE LANGE SYNDROME 1	122470		
PGM1	HGNC:8905		171900	1p31.3	NM_002633.3	Recessive	CDG1T	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IT	614921		
PHF21A	HGNC:24156		608325	11p11.2	NM_001101802.1	Dominante	IDDBCS	INTELLECTUAL DEVELOPMENTAL DISORDER WITH BEHAVIORAL ABNORMALITIES AND CRANIOFACIAL DYSMORPHISM WITH OR	618725		
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-p12.2	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		

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Gene	HGNC ID	Nom	OMIM	Locus	Transcrit RefSeq	Hérédité	Disease	Nom	OMIM	Aliases	
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		
RPL5	HGNC:10360		603634	1p22.1	NM_000969.5	Dominante	DBA6	DIAMOND-BLACKFAN ANEMIA 6	612561		
RPL11	HGNC:10301		604175	1p36.11	NM_000975.5	Dominante	DBA7	DIAMOND-BLACKFAN ANEMIA 7	612562		
RPS26	HGNC:10414		603701	12q13.2	NM_001029.5	Dominante	DBA10	DIAMOND-BLACKFAN ANEMIA 10	613309		
RPS28	HGNC:10418		603685	19p13.2	NM_001031.5	Dominante	DBA15	DIAMOND-BLACKFAN ANEMIA 15 WITH MANDIBULOFACIAL DYSOSTOSIS	606164		
SATB2	HGNC:21637		608148	2q33.1	NM_015265.3	Dominante	GLASS	GLASS SYNDROME	612313		
SETD1A	HGNC:29010		611052	16p11.2	NM_014712.2	Dominante	EPEDD	EPILEPSY, EARLY-ONSET, WITH OR WITHOUT DEVELOPMENTAL DELAY	618832		
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		
SMARCC2	HGNC:11105		601734	12q13.2	NM_001330288.2	Dominante	CSS8	COFFIN-SIRIS SYNDROME 8	618362		
SMARCD1	HGNC:11106		601735	12q13.12	NM_003076.5	Dominante	CSS11	COFFIN-SIRIS SYNDROME 11	618779		
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		
SOX4	HGNC:11200		184430	6p22.3	NM_003107.3	Dominante	CSS10	COFFIN-SIRIS SYNDROME 10	618506		
SOX9	HGNC:11204		608160	17q24.3	NM_000346.3	Dominante	CMD1; CMPD1	CAMPOMELIC DYSPLASIA	114290		
SOX11	HGNC:11191		600898	2p25.2	NM_003108.4	Dominante	CSS9	COFFIN-SIRIS SYNDROME 9	615866		
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	XLR	CPX	CLEFT PALATE WITH OR WITHOUT ANKYLOGLOSSIA, X-LINKED	303400		
TCOF1	HGNC:11654		606847	5q32-q33.1	NM_001135243.1	Dominante	TCS1	TREACHERCOLLINSSYNDROME1	154500		
TMCO1	HGNC:18188		614123	1q24.1	NM_019026.6	Recessive	CFSMR	CRANIOFACIAL DYSMORPHISM, SKELETAL ANOMALIES, AND MENTAL RETARDATION SYNDROME	213980		
TFAP2A	HGNC:11742		107580	6p24.3	NM_003220.2	Dominante	BOFS	BRANCHIOOCULOFACIALSYNDROME	113620		
TFAP2B	HGNC:11743		601601	6p12.3	NM_003221.4	Dominante	CHAR	CHAR SYNDROME	169100		
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		

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Gene	**HGNC ID**	**Nom**	**OMIM**	**Locus**	**Transcrit RefSeq**	**Hérédité**	**Disease**	**Nom**	**OMIM**	**Aliases**	
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		