

Supplementary Table S4. Autism spectrum disease (ASD)-related gene (DECIPHER).

Gene	Gene OMIM	Disease name	Disease OMIM	DDD category	Phenotypes	PMID	CES covered
ADSL	608222	ADENYLOSUCCI NASE DEFICIENCY	103050	confirmed	HP:0003196;HP:0000748;HP:0003202;HP:0005487;HP:0002179; HP:0002059;HP:0006808;HP:0000639;HP:0000248;HP:0000219; HP:0000486;HP:0002066;HP:0001336;HP:0000252;HP:0000752; HP:0003429;HP:0001348;HP:0001272;HP:0000817;HP:0000463; HP:0001249;HP:0000319;HP:0011344;HP:0001263;HP:0000718; HP:0000154;HP:0000742;HP:0001250;HP:0000717;HP:0002540; HP:0003593;HP:0000007;HP:0000343;HP:0000369;HP:0001510; HP:0000750	6150139;1 2016589;1 8830228;1 0090474;9 545543	Y
ALDH5A1	610045	SUCCINATE SEMIALDEHYDE DEHYDROGENA SE DEFICIENCY	271980	confirmed	HP:0002133;HP:0001249;HP:0100716;HP:0001251;HP:0002487; HP:0001252;HP:0001939;HP:0001265;HP:0002069;HP:0000718; HP:0000738;HP:0002121;HP:0000709;HP:0000717;HP:0003593; HP:0000007;HP:0000739;HP:0000752;HP:0000496;HP:0003812; HP:0001270;HP:0002123;HP:0002353;HP:0000750 HP:0001636;HP:0001028;HP:0004227;HP:0000175;HP:0008897; HP:0009747;HP:0002750;HP:0001511;HP:0000486;HP:0000879; HP:0000453;HP:0001631;HP:0000965;HP:0001305;HP:0006863; HP:0001388;HP:0000023;HP:0000047;HP:0008872;HP:0004322; HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028;	14635103; 9683595;1 6542398	Y
ARID1A	603024	COFFIN-SIRIS SYNDROME	135900	confirmed	HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028;	.	Y

					HP:0002650;HP:0001537;HP:0000179;HP:0000684;HP:0000776; HP:0002588;HP:0000126;HP:0001249;HP:0000280;HP:0001643; HP:0000527;HP:0000455;HP:0000151;HP:0000718;HP:0100391; HP:0002209;HP:0001629;HP:0000086;HP:0003298;HP:0000601; HP:0002808		
ASH1L	NA	INTELLECTUAL DISABILITY	NA	probable	HP:0000717;HP:0000252;HP:0004322;HP:0002650;HP:0010535	25961944; 29753921; 29276005; 28394464	Y
CAPRIN1	NA	AUTISM OR INTELLECTUAL DISABILITY	NA	possible	.	23849776	N
CHD3	NA	Macrocephaly and impaired speech and language	NA	probable	HP:0000717;HP:0000256;HP:0011398;HP:0001249;HP:0011098	30397230	N
CHD8	610528	AUTISM	209850	probable	HP:0000717;HP:0000758;HP:0011463;HP:0001249;HP:0001426; HP:0000733;HP:0002353;HP:0003144;HP:0000732;HP:0000723; HP:0003745;HP:0001250;HP:0000750;HP:0000728;HP:0000721	23160955	N
CHRNA4	NA	NOCTURNAL FRONTAL LOBE EPILEPSY TYPE 1	600513	confirmed	HP:0001249;HP:0000006;HP:0007359;HP:0003829;HP:0003621; HP:0001297;HP:0000708;HP:0001250	7647781	Y
CREBBP	600140	RUBINSTEIN- TAYBI SYNDROME TYPE 1	180849	confirmed	HP:0010775;HP:0000347;HP:0000579;HP:0000286;HP:0001763; HP:0002697;HP:0001042;HP:0008897;HP:0000481;HP:0002750; HP:0001335;HP:0002999;HP:0000486;HP:0000752;HP:0001631; HP:0000444;HP:0009765;HP:0001388;HP:0000518;HP:0000767; HP:0001371;HP:0005743;HP:0011087;HP:0009921;HP:0001956;	11331617; 7630403;2 0684013;1 2114483;1 2566391	Y

					HP:0000047;HP:0008872;HP:0005306;HP:0004322;HP:0002317; HP:0000006;HP:0000756;HP:0001274;HP:0006483;HP:0000736; HP:0001252;HP:0001425;HP:0000520;HP:0000365;HP:0000260; HP:0000160;HP:0003745;HP:0000742;HP:0001250;HP:0010562; HP:0000717;HP:0002007;HP:0001135;HP:0011675;HP:0000431; HP:0000327;HP:0000369;HP:0000490;HP:0001212;HP:0001159; HP:0009715;HP:0010055;HP:0003828;HP:0000750;HP:0000954; HP:0000278;HP:0000294;HP:0001508;HP:0001601;HP:0002880; HP:0004411;HP:0003083;HP:0000501;HP:0002162;HP:0010066; HP:0011304;HP:0001347;HP:0000574;HP:0100710;HP:0000252; HP:0000508;HP:0000077;HP:0002553;HP:0002353;HP:0000218; HP:0001007;HP:0000589;HP:0000539;HP:0000377;HP:0000028; HP:0002650;HP:0002236;HP:0002866;HP:0000957;HP:0002869; HP:0008107;HP:0005895;HP:0004209;HP:0006297;HP:0001249; HP:0002183;HP:0002870;HP:0000689;HP:0000270;HP:0002370; HP:0010442;HP:0003319;HP:0001643;HP:0002019;HP:0002144; HP:0000527;HP:0000273;HP:0002251;HP:0000678;HP:0010314; HP:0001629;HP:0000049;HP:0000494;HP:0002788;HP:0001561; HP:0000733;HP:0003298;HP:0000136;HP:0002700			
CUL3	NA	CUL3 associated autism spectrum disorder	NA	possible	HP:0000729	27824329	Y	
DEAF1	NA	Autism intellectual disability basal ganglia dysfunction and epilepsy	NA	probable	HP:0001249;HP:0000252;HP:0002134;HP:0001250	26834045; 26048982	Y	

DHCR7	602858	SMITH-LEMLI- OPITZ SYNDROME	270400	confirmed	HP:0000347;HP:0000286;HP:0000175;HP:0002827;HP:0004691; HP:0001162;HP:0000316;HP:0000048;HP:0000187;HP:0001765; HP:0001840;HP:0001830;HP:0001511;HP:0000486;HP:0000752; HP:0001631;HP:0001305;HP:0001153;HP:0001360;HP:0000518; HP:0002101;HP:0009778;HP:0000358;HP:0001622;HP:0000047; HP:0004322;HP:0000107;HP:0001845;HP:0000074;HP:0002611; HP:0007537;HP:0000365;HP:0000742;HP:0001250;HP:0009623; HP:0000717;HP:0000007;HP:0007165;HP:0000431;HP:0002013; HP:0005280;HP:0000369;HP:0000813;HP:0007333;HP:0002079; HP:0002021;HP:0010655;HP:0006979;HP:0001508;HP:0000639; HP:0001831;HP:0001276;HP:0002020;HP:0000238;HP:0000252; HP:0000171;HP:0001884;HP:0003146;HP:0000508;HP:0002566; HP:0000089;HP:0001623;HP:0000028;HP:0002089;HP:0000054; HP:0000826;HP:0000996;HP:0000126;HP:0000463;HP:0001249; HP:0001290;HP:0001643;HP:0002019;HP:0000718;HP:0000403; HP:0002033;HP:0002251;HP:0000678;HP:0000964;HP:0002579; HP:0001629;HP:0000046;HP:0001680;HP:0001558;HP:0000343; HP:0002983;HP:0000062;HP:0000104;HP:0000341;HP:0010569 HP:0010806;HP:0000722;HP:0002205;HP:0002786;HP:0000316; HP:0000232;HP:0000248;HP:0002360;HP:0002020;HP:0000158; HP:0000252;HP:0000272;HP:0010808;HP:0000741;HP:0000377; HP:0000028;HP:0000054;HP:0001762;HP:0000047;HP:0000463; HP:0001249;HP:0000006;HP:0010864;HP:0001252;HP:0001513; HP:0000303;HP:0000280;HP:0001710;HP:0000365;HP:0000718; HP:0000664;HP:0000695;HP:0003745;HP:0006335;HP:0001250; HP:0000717;HP:0000582;HP:0012210;HP:0000733;HP:0000954;	9653161;1 1857552;9 634533;12 949967;16 044199;10 677299;11 175299;12 794707;96 83613;159 52211;206 35399;108 14720;971 4007 19264732; 28498556; 16826528	Y
EHMT1	607001	Kleefstra syndrome	NA	confirmed		Y	

FMR1	NA	FRAGILE X SYNDROME	300624	confirmed	HP:0000750 HP:0000256;HP:0002003;HP:0001763;HP:0002342;HP:0003829; HP:0000303;HP:0000276;HP:0000280;HP:0002457;HP:0008640; HP:0001250;HP:0003564;HP:0000717;HP:0007165;HP:0000752; HP:0001423;HP:0001388;HP:0000767;HP:0002650;HP:0000400; HP:0002050;HP:0001634;HP:0000817	.	Y
FTSJ1	300499	MENTAL RETARDATION X-LINKED TYPE 44	309549	confirmed	HP:0000717;HP:0001256;HP:0009832;HP:0001417;HP:0005280; HP:0000750;HP:0001250;HP:0002194	10398246; 15162322; 8288232	Y
GATM	602360	ARGININE:GLYCI NE AMIDINOTRANS FERASE DEFICIENCY	612718	confirmed	HP:0000717;HP:0000007;HP:0001263;HP:0012113;HP:0000750	10762163	Y
GRIA3	305915	MENTAL RETARDATION X-LINKED TYPE 94	300699	confirmed	HP:0000256;HP:0000717;HP:0001249;HP:0001336;HP:0001533; HP:0001265;HP:0001419;HP:0002460;HP:0001250	17989220	Y
GRIN2A	138253	LANDAU- KLEFFNER SYNDROME	245570	confirmed	HP:0002353;HP:0002381;HP:0000708;HP:0010524;HP:0001250	23933818	Y
GRIN2B	138252	AUTISM MENTAL	209850	confirmed	.	23160955	Y
IL1RAPL1	300206	RETARDATION X-LINKED TYPE	300143	confirmed	HP:0000194;HP:0000582;HP:0000678;HP:0000717;HP:0001382; HP:0003196;HP:0009909;HP:0000752;HP:0002342;HP:0000316; HP:0000303;HP:0010804;HP:0000664;HP:0001419;HP:0001250	10471494; 16470793; 19012350;	Y

		21				18801879	
		MENTAL				7943039;3	
IQSEC2	300522	RETARDATION X-LINKED TYPE 1	309530	confirmed	HP:0000717;HP:0000486;HP:0000252;HP:0001249;HP:0000733; HP:0011398;HP:0002650;HP:0000248;HP:0001419;HP:0001250	177466;30 666632;20 473311	Y
KCNJ8	NA	Cantu syndrome	NA	probable	HP:0001698;HP:0001643;HP:0000280;HP:0000998;HP:0000708; HP:0001561;HP:0004948	24176758; 25275207; 24700710	Y
KDM5B	NA	Autism	NA	possible	HP:0000717;HP:0001249	28720891; 24307393	N
KDM6A	300128	KABUKI SYNDROME 2	300867	confirmed	HP:0002000;HP:0008872;HP:0000637;HP:0004322;HP:0001263; HP:0007655;HP:0000689;HP:0001998;HP:0011398;HP:0000437; HP:0000527;HP:0000455;HP:0000486;HP:0000252;HP:0000769; HP:0001423;HP:0001680;HP:0000378;HP:0002553;HP:0002761; HP:0000668;HP:0000411;HP:0001212;HP:0001631;HP:0001007; HP:0000218;HP:0001156;HP:0004325;HP:0000708;HP:0005338	22197486; 23076834	Y
KMT2C	NA	INTELLECTUAL DISABILITY	NA	probable	HP:0000717;HP:0001249	29276005; 29069077	N
KMT2E	NA	INTELLECTUAL DISABILITY	NA	confirmed	HP:0000717;HP:0000256;HP:0001249;HP:0001250;HP:0000490; HP:0002808	.	N
MAGEL2	NA	Schaaf-Yang syndrome	NA	confirmed	HP:0000717;HP:0030044;HP:0001263	27195816; 24076603	Y
MBOAT7	NA	Intellectual Disability Accompanied by Epilepsy and	NA	probable	HP:0000717;HP:0001249;HP:0001250	27616480	N

Autistic Features							
MED12	300188	LUJAN-FRYNS SYNDROME	309520	confirmed	HP:0000194;HP:0000256;HP:0000347;HP:0000722;HP:0001547; HP:0000735;HP:0001611;HP:0003189;HP:0000426;HP:0011220; HP:0011304;HP:0000119;HP:0000219;HP:0000712;HP:0000752; HP:0000275;HP:0001631;HP:0000218;HP:0001388;HP:0000767; HP:0001371;HP:0001166;HP:0002631;HP:0000322;HP:0001249; HP:0000446;HP:0001274;HP:0001519;HP:0008544;HP:0001290; HP:0000276;HP:0000718;HP:0000744;HP:0001250;HP:0000717; HP:0000678;HP:0000709;HP:0002007;HP:0001629;HP:0000327; HP:0002002;HP:0000369;HP:0001419	6711603	Y
MED13	NA	MED13 - Neurodevelopment disorder	NA	probable	HP:0001249;HP:0000729;HP:0001263;HP:0000750	29740699	N
MTHFR	NA	METHYLENETET RAHYDROFOLAT E REDUCTASE DEFICIENCY	236250	confirmed	HP:0000007;HP:0002160;HP:0000252;HP:0002156;HP:0001263; HP:0001324;HP:0001288;HP:0002311;HP:0003401;HP:0001297; HP:0000708;HP:0001250	.	Y
NBEA	NA	NBEA Neurodevelopment disorder with seizures	NA	probable	HP:0001249;HP:0011170;HP:0000729;HP:0010819;HP:0001263; MESH:D004827	30269351	N
NEXMIF	NA	KIAA2022	300912	probable	HP:0000717;HP:0000486;HP:0001249	15466006; 23615299	N
NFIB	NA	Intellectual disability with macrocephaly	NA	probable	HP:0000256;HP:0001256;HP:0000729	30388402	N

NHS	300457	NANCE-HORAN SYNDROME	302350	confirmed	HP:0000482;HP:0002342;HP:0000699;HP:0000568;HP:0000426;HP:0009803;HP:0000639;HP:0000501;HP:0000276;HP:0006346;HP:0000448;HP:0000717;HP:0001500;HP:0008031;HP:0000275;HP:0001423;HP:0000572;HP:0000519;HP:0000400;HP:0006332	458526;15 623749;14 564667;22 46772	Y
NLGN3	300336	AUTISM SPECTRUM DISORDERS	198890	probable	.	12669065	Y
NLGN4X	NA	SUSCEPTIBILITY TO AUTISM X-LINKED TYPE 2	300495	possible	HP:0001249;HP:0001426;HP:0002332;HP:0000732;HP:0000723;HP:0003745;HP:0001250;HP:0000717;HP:0000758;HP:0011463;HP:0005324;HP:0001417;HP:0000733;HP:0002353;HP:0003144;HP:0000721;HP:0000750	.	Y
NRXN1	600565	AUTISM	209850	probable	HP:0000717;HP:0000758;HP:0011463;HP:0001249;HP:0001426;HP:0000733;HP:0002353;HP:0003144;HP:0000732;HP:0000723;HP:0003745;HP:0001250;HP:0000750;HP:0000728;HP:0000721	.	Y
NRXN2	NA	AUTISM	209850	probable	HP:0000717;HP:0000758;HP:0011463;HP:0001249;HP:0001426;HP:0000733;HP:0002353;HP:0003144;HP:0000732;HP:0000723;HP:0003745;HP:0001250;HP:0000750;HP:0000728;HP:0000721	.	N
NRXN3	NA	AUTISM	209850	possible	HP:0000717;HP:0000758;HP:0011463;HP:0001249;HP:0001426;HP:0000733;HP:0002353;HP:0003144;HP:0000732;HP:0000723;HP:0003745;HP:0001250;HP:0000750;HP:0000728;HP:0000721	22209245	N
NSD1	606681	WEAVER SYNDROME	277590	confirmed	HP:0000256;HP:0000278;HP:0000034;HP:0000286;HP:0006956;HP:0001331;HP:0001608;HP:0002673;HP:0000316;HP:0011304;HP:0001840;HP:0000486;HP:0001176;HP:0009466;HP:0003066;HP:0000973;HP:0001350;HP:0001761;HP:0000028;HP:0002650;HP:0000400;HP:0001814;HP:0200000;HP:0001537;HP:0002866;HP:0000708;HP:0001762;HP:0004689;HP:0003186;HP:0000023;	.	Y

NSD1	606681	SOTOS SYNDROME	117550	confirmed	HP:0001249;HP:0000006;HP:0001848;HP:0001263;HP:0000773; HP:0001845;HP:0008070;HP:0003015;HP:0001252;HP:0000303; HP:0005616;HP:0001257;HP:0001250;HP:0001377;HP:0010751; HP:0000494;HP:0009473;HP:0005280;HP:0000343;HP:0001540; HP:0001212;HP:0000311;HP:0001260;HP:0000750;HP:0002808 HP:0000256;HP:0000098;HP:0001763;HP:0000307;HP:0001792; HP:0001338;HP:0001319;HP:0000316;HP:0000405;HP:0000639; HP:0002280;HP:0006288;HP:0001347;HP:0002389;HP:0000486; HP:0001176;HP:0000388;HP:0001631;HP:0000218;HP:0001388; HP:0002650;HP:0002667;HP:0000708;HP:0002474;HP:0000268; HP:0000006;HP:0001833;HP:0001263;HP:0002370;HP:0005616; HP:0000303;HP:0001643;HP:0003745;HP:0001250;HP:0002857; HP:0002007;HP:0000540;HP:0001629;HP:0000494;HP:0009890; HP:0001952	12525543; 16222665; 11896389	Y
OTUD6B	NA	Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features	NA	probable	HP:0000717;HP:0000252;HP:0001249;HP:0001263;HP:0011968; HP:0001257;HP:0003510	28343629	N
OTUD7A	NA	15q13.3 deletions phenocopy	NA	possible	HP:0000717;HP:0001249;HP:0001250	29395074	N
PRR12	NA	Intellectual disability and iris abnormalities	NA	probable	HP:0000589;HP:0001249;HP:0000729;HP:0001263;HP:0012775	29556724; 26163108	N

PTCHD1	NA	AUTISM/ID MACROCEPHAL	300830	confirmed	.	20844286	Y
PTEN	601728	Y/AUTISM SYNDROME Intellectual Disability with Speech Delay	605309	confirmed	HP:0000717;HP:0002007;HP:0003196;HP:0000006;HP:0005490; HP:0001263;HP:0004422;HP:0005280;HP:0000343;HP:0001513; HP:0000337	17286265; 23160955; 15805158	Y
PUS7	NA	Microcephaly Short Stature and Aggressive Behavior	NA	probable	.	30526862	N
SHANK1	NA	AUTISM	209850	probable	HP:0000717;HP:0000758;HP:0011463;HP:0001249;HP:0001426; HP:0000733;HP:0002353;HP:0003144;HP:0000732;HP:0000723; HP:0003745;HP:0001250;HP:0000750;HP:0000728;HP:0000721	.	N
SHANK2	NA	SUSCEPTIBILITY TO AUTISM TYPE 17	613436	probable	.	20473310	Y
SHANK3	606230	PHELAN- MCDERMID SYNDROME	606232	probable	HP:0000256;HP:0100704;HP:0000076;HP:0000286;HP:0100797; HP:0004691;HP:0002119;HP:0001004;HP:0000486;HP:0000113; HP:0000272;HP:0000966;HP:0011968;HP:0000400;HP:0000268; HP:0002046;HP:0000293;HP:0002317;HP:0002572;HP:0000164; HP:0001800;HP:0011120;HP:0007328;HP:0000365;HP:0003745; HP:0001250;HP:0000717;HP:0000431;HP:0100658;HP:0000490; HP:0002136;HP:0002518;HP:0003763;HP:0000960;HP:0000750; HP:0000098;HP:0100540;HP:0002342;HP:0000307;HP:0001319; HP:0002188;HP:0000574;HP:0002020;HP:0000252;HP:0001176;	22892527; 17173049	Y

					HP:0000508;HP:0001270;HP:0000218;HP:0000817;HP:0000414; HP:0004209;HP:0001263;HP:0000689;HP:0001290;HP:0001265; HP:0100702;HP:0000710;HP:0001643;HP:0000527;HP:0000718; HP:0000331;HP:0001629;HP:0000343;HP:0000411;HP:0100703		
SIM1	NA	Severe obesity with neurobehavioral features	NA	confirmed	HP:0006889;HP:0000729;HP:0000736;HP:0008915	23778139; 28472148; 23778136	Y
SLC9A9	NA	SUSCEPTIBILITY TO AUTISM TYPE 16	613410	possible	.	.	N
SMARCA 2	600014	COFFIN SIRIS	135900	confirmed	HP:0001636;HP:0001028;HP:0004227;HP:0000175;HP:0008897; HP:0009747;HP:0002750;HP:0001511;HP:0000486;HP:0000879; HP:0000453;HP:0001631;HP:0000965;HP:0001305;HP:0006863; HP:0001388;HP:0000023;HP:0000047;HP:0008872;HP:0004322; HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028; HP:0002650;HP:0001537;HP:0000179;HP:0000684;HP:0000776; HP:0002588;HP:0000126;HP:0001249;HP:0000280;HP:0001643; HP:0000527;HP:0000455;HP:0000151;HP:0000718;HP:0100391; HP:0002209;HP:0001629;HP:0000086;HP:0003298;HP:0000601; HP:0002808	22426308	Y
SMARCA	603254	COFFIN SIRIS	135900	confirmed	HP:0001636;HP:0001028;HP:0004227;HP:0000175;HP:0008897;	.	Y

4					HP:0009747;HP:0002750;HP:0001511;HP:0000486;HP:0000879; HP:0000453;HP:0001631;HP:0000965;HP:0001305;HP:0006863; HP:0001388;HP:0000023;HP:0000047;HP:0008872;HP:0004322; HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028; HP:0002650;HP:0001537;HP:0000179;HP:0000684;HP:0000776; HP:0002588;HP:0000126;HP:0001249;HP:0000280;HP:0001643; HP:0000527;HP:0000455;HP:0000151;HP:0000718;HP:0100391; HP:0002209;HP:0001629;HP:0000086;HP:0003298;HP:0000601; HP:0002808 HP:0001636;HP:0001028;HP:0004227;HP:0000175;HP:0008897; HP:0009747;HP:0002750;HP:0001511;HP:0000486;HP:0000879; HP:0000453;HP:0001631;HP:0000965;HP:0001305;HP:0006863; HP:0001388;HP:0000023;HP:0000047;HP:0008872;HP:0004322; HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028; HP:0002650;HP:0001537;HP:0000179;HP:0000684;HP:0000776; HP:0002588;HP:0000126;HP:0001249;HP:0200022;HP:0000280;	25249037; 23929686; 22726846; 23906836; 23815551; 29907796; 22426308	Y
SMARCB1	601607	EHMT1-like SYNDROME	NA	confirmed			

SMARCE1	603111	COFFIN SIRIS	135900	probable	HP:0001643;HP:0000527;HP:0000455;HP:0000151;HP:0000718; HP:0100391;HP:0002209;HP:0001629;HP:0000086;HP:0003298; HP:0000601;HP:0002808 HP:0001636;HP:0001028;HP:0004227;HP:0000175;HP:0008897; HP:0009747;HP:0002750;HP:0001511;HP:0000486;HP:0000879; HP:0000453;HP:0001631;HP:0000965;HP:0001305;HP:0006863; HP:0001388;HP:0000023;HP:0000047;HP:0008872;HP:0004322; HP:0001252;HP:0008398;HP:0000365;HP:0000154;HP:0001250; HP:0000007;HP:0002576;HP:0005280;HP:0002079;HP:0000729; HP:0006498;HP:0000954;HP:0000960;HP:0002205;HP:0002219; HP:0002673;HP:0001338;HP:0003083;HP:0000639;HP:0000483; HP:0000574;HP:0000252;HP:0000545;HP:0000384;HP:0000508; HP:0002566;HP:0000089;HP:0002592;HP:0000218;HP:0000028; HP:0002650;HP:0001537;HP:0000179;HP:0000684;HP:0000776; HP:0002588;HP:0000126;HP:0001249;HP:0000280;HP:0001643; HP:0000527;HP:0000455;HP:0000151;HP:0000718;HP:0100391; HP:0002209;HP:0001629;HP:0000086;HP:0003298;HP:0000601; HP:0002808	22426308	Y
SRP54	NA	Syndromic neutropenia with Shwachman-Diamond-like features	NA	probable	HP:0001875;HP:0001738;HP:0000729	28972538	N
TBL1XR1	NA	Intellectual disability with autism spectrum	NA	confirmed	HP:0001249;HP:0001426;HP:0000939;HP:0000732;HP:0000723; HP:0003745;HP:0001250;HP:0000728;HP:0000717;HP:0011463; HP:0000758;HP:0000733;HP:0002353;HP:0001156;HP:0003144;	25425123; 23160955	N

TBR1	NA	disorder AUTISM	209850	probable	HP:0000721;HP:0000750	23160955	N
TRIP12	NA	TRIP12-related intellectual disability	NA	confirmed	HP:0001249;HP:0000729	27848077; 28251352	N
TRRAP	NA	with/without autism spectrum disorder Autism and Syndromic Intellectual Disability	NA	probable	.	30827496	N
TSC1	605284	TUBEROUS SCLEROSIS TYPE 1	191100	confirmed	HP:0009720;HP:0009554;HP:0005584;HP:0010762;HP:0001328; HP:0200024;HP:0011097;HP:0009724;HP:0009722;HP:0009719; HP:0000169;HP:0003812;HP:0009727;HP:0009716;HP:0000957; HP:0000826;HP:0002514;HP:0007018;HP:0009734;HP:0001249; HP:0000006;HP:0001482;HP:0000107;HP:0006772;HP:0009721; HP:0000821;HP:0009729;HP:0001716;HP:0009592;HP:0009717; HP:0000717;HP:0002086;HP:0002888	18830229; 10053179; 10340649; 9242607	Y
TSC2	191092	TUBEROUS SCLEROSIS TYPE 2	613254	confirmed	HP:0002514;HP:0007018;HP:0009734;HP:0009720;HP:0001249; HP:0000006;HP:0001482;HP:0000107;HP:0005584;HP:0006772; HP:0009721;HP:0010762;HP:0000821;HP:0001328;HP:0009729; HP:0001716;HP:0009592;HP:0009717;HP:0011097;HP:0009724; HP:0000717;HP:0000169;HP:0003812;HP:0009727;HP:0002888; HP:0009716;HP:0000957;HP:0000826	7581393;1 0069705;1 7120248;1 9259131;1 0206124;9 361032;93 02281;882 5048;9463	Y

						313;88248	
						81;127525	
						78;863470	
						1;1140304	
						7	
		MENTAL RETARDATION					
ZDHHC9	300646	SYNDROMIC X- LINKED	300799	confirmed	HP:0000486;HP:0001249;HP:0009183;HP:0001763;HP:0001417; HP:0001519;HP:0000411;HP:0000768;HP:0001166;HP:0000708	17436253; 26000327	Y
		ZDHHC9- RELATED					
ZNF713	NA	AUTISM	209850	possible	.	25196122	N

Y: Covered by CES target; N: not covered by CES target.