Table 1: Pathogenic/likely pathogenic variants and Variants of Uncertain Significance (VUS) detected with WES in genes associated with referral conditions Variant classification according to ACMG guidelines.

Gene	Case ID/ Gender	Age (yrs) at testing	Variant	Zygosity	Inheritance mode	Segregation	MIM Disease (MIM number)	Novel/ Known	Classification (ACMG score)
Neurodevelopmental abnormalities									
ACTB	8056/F	4	NM_001101.4:c.547C>T, p.(Arg183Trp)	Het	AD	De novo	Baraitser-Winter syndrome 1 (243310)	Known	Pathogenic (PS2, PM2, PP2, PP3, PP4, PP5)
ACTL6B	9107/M	2	NM_016188.5:c.1027G>A, p.(Gly343Arg)	Het	AD	De novo	Intellectual developmental disorder with severe speech and ambulation defects (618470)	Known	Pathogenic (PS2, PS3, PM2, PM6, PP3, PP4, PP5)
ATP7A	9102/M	1	NM_000052.7:c.2383C>T, p.(Arg795*)	Hem	XLR	Mat	Menkes disease (309400)	Known	Pathogenic (PVS1, PM1, PM2, PP3, PP5)
CIC	8051/M	7	NM_015125.4:c.436_437delAG, p.(Ser146*)	Het	AD	De novo	Mental retardation, autosomal dominant 45 (617600)	Novel	Pathogenic (PVS1, PS2, PM2, PP3)
DDX3X	8109/F	29	NM_001193417.2:c.883C>T, p.(Arg295*)	Het	XLD/XLR	n/a	Mental retardation, X-linked 102 (300958)	Novel	Pathogenic (PVS1, PM1, PM2, PP3)
DLG3	8068/M	7	NM_021120.4:c.251C>T, p.(Pro84Leu)	Het	XLR	Mat	Mental retardation, X-linked 90 (300850)	Novel	Likely Pathogenic (PM2, PP3, PP4)
GABBR2	8137/F	11	NM_005458.8:c.2119G>A, p.(Ala707Thr)	Het	AD	De novo	Epileptic encephalopathy, early infantile, 59 (617904)	Novel	Likely Pathogenic (PS2, PM2, PP3, PP4)
GRIN2B	9124/F	5	NM_000834.4:c.1606G>A, p.(Val536Ile)	Het	AD	De novo	Mental retardation, autosomal dominant 6 (613970)	Novel	Pathogenic (PS2, PM1, PM2, PP2, PP3)
ITPR1	9032/M	17	NM_001168272.1:c.748T>C, p.(Phe250Leu)	Het	AD	De novo	Spinocerebellar ataxia 15 (606658)	Novel	Likely Pathogenic (PS2, PM1, PM2, PP3)
ITSN1	9090/M	23	NM_003024.3: c.2842_2843delAT,	Het	AD	n/a	Autistic spectrum disorder (n/a)	Novel	Likely Pathogenic (PVS1, PM2, PP3)
KDM6A	8074/M	8	NM_021140.3:c.2645_2646insAC, p.(Pro883Hisfs*16)	Het	XLD	De novo	Kabuki syndrome 2 (300867)	Novel	Pathogenic (PVS1, PS2, PM1, PM2, PP3, PP4)

MAGEL2	8023/M	3	NM_019066.4:c.1996dupC, p.(Gln666Profs*47)	Het	AD	De novo	Schaaf-Yang syndrome (615547)	Known	Pathogenic (PVS1, PS2, PM2, PP3, PP5)
MECP2	8080/F	30	NM_004992.3:c.916C>T, p.(Arg306Cys)	Het	AD	De novo	Rett syndrome (312750)	Known	Pathogenic (PS2, PM1, PM2, PM5, PP2, PP3, PP4, PP5)
MED12	8009/M	15	NM_005120.2:c.4021C>T, p.(Arg1341Trp)	Hem	XLR	n/a	Lujan-Fryns syndrome (309520)	Novel	Likely Pathogenic (PM2, PP2, PP3, PP4)
MKS1	8125/M	3	NM_017777.3:c.370C>T, p.(Arg124*)/c.1476T>G, p.(Cys492Trp)	Comp Het	AD	Mat/Pat	Joubert syndrome 28 (617121)	Novel/ Known	Pathogenic/Likely Pathogenic (PVS1, PM2, PM3, PP3/PM2, PP2, PP3, PP5)
NALCN	8005/F	3	NM_052867.4:c.5209G>C, p.(Asp1737His)	Het	AD	De novo	Congenital contractures of the limbs and face, hypotonia, and developmental delay (616266)	Novel	Likely Pathogenic (PS2, PM2, PP2, PP3)
PURA	8070/F	2	NM_005859.4:c.224T>G, p.(Leu75Arg)	Het	AD	De novo	Mental retardation, autosomal dominant 31 (616158)	Novel	Likely Pathogenic (PM1, PM2, PM5, PP2, PP3)
SET	9151/F	9	NM_001248000.2:c.99_102delAGAC, p.(Arg35Leufs*10)	Het	AD	n/a	Mental retardation, autosomal dominant 58 (618106)	Novel	Pathogenic (PVS1, PM2, PP3)
STXBP1	9103/M	2	NM_003165.4:c.993delG, p.(Lys332Argfs*24)	Het	AD	n/a	Epileptic encephalopathy, early infantile, 4 (612164)	Novel	Likely Pathogenic (PVS1, PM2, PP3)
TGM6	8091/F	5	NM_198994.2:c.1336+1G>T	Het	AR	Pat (same phenotype)	Spinocerebellar ataxia 35 (613908)	Novel	Pathogenic (PVS1, PM2, PP3)
Skeletal/connective tissue abnormalities									
CHRNG	9050/F	7	NM_005199.5:c.715C>T, p.(Arg239Cys)/c.895T>A, p.(Ser299Thr)	Comp Het	AR	Mat/Pat	Escobar syndrome (265000)	Known/ Novel	Pathogenic/Likely Pathogenic (PS3, PM2, PP2, PP3, PP5/PM2, PM3, PP2, PP3, PP4)
COL11A1	8010/M	8	NM_001854.4:c.3427A>C, p.(Lys1143Gln)	Het	AD	n/a	Stickler syndrome, type II (604841)	Novel	Likely Pathogenic (PM1, PM2, PP2, PP3, PP4)
COL1A1	8114/M	2	NM_000088.3:c.3494delC, p.(Pro1164Leufs*74)	Het	AD	n/a	Osteogenesis imperfecta, type I (166200)	Novel	Pathogenic (PVS1, PM1, PM2, PP3, PP4)

COL2A1	8053/F	37	NM_001844.4:c.3136C>T, p.(Pro1046Ser)	Het	AD	n/a	Osteoarthritis with mild chondrodysplasia (604864)	Novel	Likely Pathogenic (PM1, PM2, PP2, PP3)
COL5A1	8075/M	5	NM_001278074.1:c.3751dupC, p.(Pro1253Serfs*14)	Het	AD	n/a	Ehlers-Danlos syndrome, classic type, 1 (130000)	Known	Pathogenic (PVS1, PM1, PM2, PP4)
FBN1	8117/F	33	NM_000138.4:c.1669T>C, p.(Cys557Arg)	Het	AD	n/a	Marfan syndrome (154700)	Novel	Likely Pathogenic (PM1, PM2, PM5, PP2, PP3, PP4)
FBN1	9057/M	5	NM_000138.4:c.7454A>G, p.(Asp2485Gly)	Het	AD	n/a	Marfan syndrome (154700)	Known	Likely Pathogenic (PM1, PM2, PP3, PP2, PP4, PP5)
FBN1	9089/F	31	NM_000138.4:c.6380A>G, p.(Asp2127Gly)	Het	AD	n/a	Marfan syndrome (154700)	Known	Likely Pathogenic (PM1, PM2, PM5, PP3)
FBN1	9125/M	24	NM_000138.4:c.7712G>A, p.(Cys2571Tyr)	Het	AD	n/a	Marfan syndrome (154700)	Known	Pathogenic (PM1, PM2, PM5, PP2, PP3, PP4, PP5)
FBN1	9150/F	4	NM_000138.4:c.5076_5078delAAG, p.(Arg1692del)	Het	AD	n/a	Marfan syndrome (154700)	Novel	Likely Pathogenic (PM1, PM2, PM4, PP3)
GALNS	9144/M	8	NM_000512.5:c.719A>G, p.(Tyr240Cys)	Hom	AR	n/a	Mucopolysaccharidosis IVA (253000)	Known	Pathogenic (PM1, PM2, PP2, PP3, PP5)
GNAS	8108/M	14	NM_080425.3:c.2293C>G, p.(Prp765Ala)	Het	AD	n/a	Pseudohypoparathyroidism Ia/b/c (103580/603233/612462)	Novel	Likely Pathogenic (PM1, PM2, PP2, PP3)
KIF22	8113/M	22	NM_007317.2:c.1891-3C>G	Het	AD	De novo	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 (603546)	Novel	Likely Pathogenic (PS2, PM2, PP3)
NOG	8135/F	6	NM_005450.4:c.379G>T, p.(Glu127*)	Het	AD	De novo	Tarsal-carpal coalition syndrome (186570)	Novel	Pathogenic (PVS1, PS2, PM2, PP3)
PRDM5	9128/M	22	NM_018699.3:c.974delG, p.(Cys325Leufs*2)	Hom	AR	n/a	Brittle cornea syndrome 2 (614170)	Known	Pathogenic (PVS1, PM2, PP3, PP5)
TGFB2	9058/M	25	NM_001135599.3:c.996_1014del, p.(Asp333Glufs*20)	Het	AD	n/a	Loeys-Dietz syndrome 4 (614816)	Novel	Pathogenic (PVS1, PM1, PM2, PP3)
WISP3	8076/F	14	NM_198239.1:c.643G>C, p.(Ala215Pro)	Hom	AR	Mat/Pat	Spondyloepiphyseal dysplasia tarda with progressive arthropathy/Arthropathy, progressive pseudorheumatoid, of childhood (208230)	Novel	Likely Pathogenic (PM2, PM3, PP3, PP4)

Congenital anomalies/syndromic									
DDX59	8006/M	9	NM_001031725.6:c.751T>C, p.(Ser251Pro)/c.1597-6T>G	Comp Het	AR	Mat/Pat	Orofaciodigital syndrome V (174300)	Novel/ Novel	Likely Pathogenic/Likely Pathogenic (PM2, PP3, PP4/PM2, PM3, PP3, PP4)
KAT6B	8008/F	3	NM_012330.4:c.5167dupC, p.(Gln1723Profs*52)	Het	AD	n/a	SBBYSS syndrome (603736)	Novel	Pathogenic (PVS1, PM1, PM2, PP3, PP4)
KRAS	8007/F	6	NM_033360.3:c.458A>T, p.(Asp153Val)	Het	AD	n/a	Noonan syndrome 3 (609942)	Known	Likely Pathogenic (PS3, PM2, PP2, PP3, PP5)
NSD1	9118/F	1	NM_022455.4:c.5824_5828delCCAGA, p.(Pro1942Glyfs*2)	Het	AD	De novo	Sotos syndrome 1 (117550)	Novel	Pathogenic (PVS1, PM1, PM2, PP3)
ORC4	9070/M	3	NM_181742.3:c.623C>G, p.(Ser208*)/c.956A>G, p.(His319Arg)	Comp Het	AR	Pat/Mat	Meier-Gorlin syndrome 2 (613800)	Novel/ Novel	Pathogenic/Likely Pathogenic (PVS1, PM2, PP3/PM2, PM3, PP3, PP4)
PIK3R1	9127/M	3	NM_181523.3:c.1945C>T, p.(Arg649Trp)	Het	AD	De novo	SHORT syndrome (269880)	Known	Pathogenic (PS2, PS3, PM1, PM2, PP3, PP5)
PTCH1	8111/M	18	NM_000264.4:c.1196delG, p.(Trp399Cysfs*33)	Het	AD	n/a	Basal cell nevus syndrome (109400)	Novel	Pathogenic (PVS1, PM2, PP3, PP4)
PTPN11	8112/M	5	NM_002834.4:c.922A>G, p.(Asn308Asp)	Het	AD	n/a	Noonan syndrome 1 (163950)	Known	Pathogenic (PS3, PM1, PM2, PM5, PP2, PP3, PP4, PP5)
PTPN11	8133/F	23	NM_002834.4:c.188A>G, p.(Tyr63Gly)	Het	AD	n/a	Noonan syndrome 1 (163950)	Known	Pathogenic (PS3, PM1, PM2, PP2, PP3, PP5)
PTPN11	9080/M	2	NM_002834.4:c.236A>G, p.(Gln79Arg)	Het	AD	n/a	Noonan syndrome 1 (163950)	Known	Pathogenic (PM1, PM2, PM5, PP3, PP4, PP5)
PTPN11	9138/F	14	NM_002834.4:c.922A>G, p.(Gly503Arg)	Het	AD	n/a	Noonan syndrome 1 (163950)	Known	Pathogenic (PM1, PM2, PM5, PP2, PP3, PP5)
PTPN11	9148/F	17	NM_002834.4:c.1507G>C, p.(Gly503Arg)	Het	AD	n/a	Noonan syndrome 1 (163950)	Known	Pathogenic (PS1, PM1, PM2, PM5, PP2, PP3, PP5)
PTPN11	9165/M	3	NM_002834.4:c.1381G>T, p.(Ala461Ser)	Het	AD	n/a	Leopard syndrome 1 (151100)	Known	Pathogenic (PM1, PM2, PM5, PP2, PP3, PP5)

RIT1	9030/F	2	NM_006912.6:c.270G>A, p.(Met90Ile)	Het	AD	De novo	Noonan syndrome 8 (615355)	Known	Pathogenic (PS1, PS2, PM1, PM2, PM5, PP2, PP3, PP4, PP5)
RIT1	9044/F	2	NM_006912.6:c.244T>G, p.(Phe82Val)	Het	AD	De novo	Noonan syndrome 8 (615355)	Known	Pathogenic (PS2, PM1, PM2, PM5, PP2, PP3, PP4, PP5)
SHOC2	9106/F	1	NM_007373.3:c.4A>G, p.(Ser2Gly)	Het	AD	De novo	Noonan syndrome-like with loose anagen hair (607721)	Known	Pathogenic (PS2, PM2, PP3, PP5)
Neuromuscular abnormalities									
ANO5	8017/M	51	NM_001142649.1:c.1517delT, p.(Phe506Serfs*6)/Exon 5 Deletion	Comp Het	AR	Mat/Pat	Miyoshi muscular dystrophy 3/Muscular dystrophy, limb-girdle 12 (613319/611307)	Known/ Novel	Pathogenic/Pathogenic (PVS1, PM2, PP3, PP5/PVS1, PM2, PP3)
CACNA1S	9056/M	22	NM_000069.3:c.2627T>A, p.(Val876Glu)	Het	AD	n/a	Hypokalemic periodic paralysis, type 1 (170400)	Known	Likely Pathogenic (PM2, PP2, PP3, PP4, PP5)
CAPN3	9059/M	5	NM_000070.2:c.1611C>A, p.(Tyr537*)	Hom	AR	Mat/Pat	Muscular dystrophy, limb-girdle, autosomal recessive 1 (253600)	Known	Pathogenic (PVS1, PM1, PM2, PP3, PP5)
CLCN1	9028/F	35	NM_000083.2:c.2680C>T, p.(Arg894*)	Hom	AR	n/a	Myotonia congenita, recessive (255700)	Known	Pathogenic (PVS1, PM1, PM2, PP3)
COL6A1	9020/F	38	NM_001848.2:c.850G>A, p.(Gly284Arg)	Het	AD	n/a	Bethlem myopathy 1/Ullrich congenital muscular dystrophy 1 (158810/254090)	Known	Pathogenic (PS1, PM1, PM2, PP2, PP3, PP5)
DNM2	9116/F	2	NM_001005361.2:c.1853C>A, p.(Ala618Asp)	Het	AD	De novo	Centronuclear myopathy 1 (160150)	Known	Pathogenic (PS2, PM1, PM2, PM5, PP2, PP3, PP5)
LMNA	8057/M	5	NM_170707.3:c.745C>T, p.(Arg249Trp)	Het	AD	De novo	Muscular dystrophy, congenital (613205)	Known	Likely Pathogenic (PS2, PM1, PM2, PM5, PP2, PP3, PP5)
RYR1	8138/F	3	NM_000540.2:c.1250T>C, p.(Leu417Pro)/c.1264G>A, p.(Gly422Arg)	Comp Het	AR	Pat/Mat	Minicore myopathy with external ophthalmoplegia (255320)	Known/ Known	Likely Pathogenic/Likely Pathogenic (PM2, PM3, PP2, PP3, PP5/PM2, PM3, PP2, PP3)

RYR1	9051/F	8	NM_000540.2:c.1250T>C, p.(Leu417Pro)/c.6445G>A, p.(Val2149Met)	Comp Het	AR	Mat/Pat	Minicore myopathy with external ophthalmoplegia (255320)	Known/ Novel	Likely Pathogenic/Likely Pathogenic (PM2, PP2, PP3, PP5/PM1, PM2, PP2, PP3, PP4)
SPAST	9115/F	3	NM_014946.3:c.1245+5G>A/ c.131C>T, p.(Ser44Leu)	Comp Het	AR	Pat/Mat	Spastic paraplegia 4, autosomal dominant (182601)	Known/ Known	Pathogenic/Modifier polymorphism (PM1, PM2, PP3, PP4, PP5)
Ocular/Auditory abnormalities									
GJB2	8016/M	9	NM_004004.5:c.71G>A, p.(Trp24*)	Hom	AR	n/a	Deafness, autosomal recessive 1A (220290)	Known	Pathogenic (PVS1, PM1, PP3, PP5)
GJB2	8039/F	10	NM_004004.5:c.35delG, p.(Gly12Valfs*2)	Hom	AR	n/a	Deafness, autosomal recessive 1A (220290)	Known	Pathogenic (PVS1, PS3, PM1, PP5)
COL4A1	8011/F	3	NM_001845.5:c.2185G>C, p.(Gly729Arg)	Het	AD	De novo	Brain small vessel disease with or without ocular anomalies (175780)	Novel	Likely Pathogenic (PS2, PM1, PM2, PP2, PP3)
GUCY2D	8139/F	25	NM_000180.3:c.2302C>T, p.(Arg768Trp)	Hom	AR	n/a	Leber congenital amaurosis 1 (204000)	Known	Pathogenic (PM5, PM1, PP2, PP3, PP4, PP5)
RP2	9104/F	14	NM_006915.3:c.969+3A>C	Hem	XLR	Mat	Retinitis pigmentosa 2 (312600)	Known	Likely Pathogenic (PM2, PP3, PP4, PP5)
Cardio and/or vascular abnormalities									
ENG	9062/F	4	NM_001114753.2:c.296T>A, p.(Leu99His)	Het	AD	Pat (same phenotype)	Telangiectasia, hereditary hemorrhagic, type 1 (187300)	Novel	Likely Pathogenic (PM1, PM2, PP1, PP2, PP3, PP4)
RNF213	8073/F	20	NM_001256071.2:c.12059G>T, p.(Cys4020Phe)	Het	AD	Mat (same phenotype)	Moyamoya disease 2 (607151)	Novel	Likely Pathogenic (PM1, PM2, PP1, PP3, PP4)
Dermatological abnormalities									
ALOX12B	9113/M	1	NM_001139.3:c.805C>T, p.(Leu269Phe)/c.1562A>G, p.(Tyr521Cys)	Comp Het	AR	Pat/Mat	Ichthyosis, congenital, autosomal recessive 2 (242100)	Novel/ Known	Likely Pathogenic/Pathogenic (PM1, PM2, PM3, PP2, PP3/PM1, PM2, PP2, PP3, PP4, PP5)
FERMT1	9073/F	2	NM_017671.4:c.676dupC, p.(Gln226Profs*17)	Hom	AR	Mat/Pat	Kindler syndrome (173650)	Known	Pathogenic (PVS1, PM1, PM2, PP5)
GJB2	9088/M	3	NM_004004.6:c.524C>G, p.(Pro175Arg)	Het	AD	n/a	Vohwinkel syndrome (124500)	Novel	Likely Pathogenic (PM1, PM2, PP2, PP3)

KLHL24	8052/M	3	NM_017644.3:c.1A>G, p.(Met1Val)	Het	AD	n/a	Epidermolysis bullosa simplex, generalized, with scarring and hair loss (617294)	Known	Pathogenic (PVS1, PM2, PP3, PP4, PP5)
KRT10	9019/M	3	NM_000421.1:c.449T>C, p.(Met150Thr)	Het	AD	n/a	Ichthyosis, cyclic, with epidermolytic hyperkeratosis (607602)	Known	Pathogenic (PM1, PM2, PM5, PP2, PP3, PP4, PP5)
Metabolism abnormalities									
GFAP	7014/M	3	NM_002055.4:c.242C>A, p.(Ala81Asp)	Het	AD	De novo	Alexander disease (203450)	Novel	Pathogenic (PS2, PM1, PM2, PP2, PP3)
GNPTAB	8110/M	8	NM_024312.4:c.569A>T, p.(Asp190Val)/c.2868_2869delCA, p.(Met957Alafs*5)	Comp Het	AR	Mat/Pat	Mucolipidosis II/III alpha/beta (252500/252600)	Known/ Novel	Likely Pathogenic/Pathogenic (PM2, PP2, PP3, PP4, PP5/PVS1, PM2, PP3, PP4)
PDHA1	8030/M	4	NM_001173454.1:c.863C>T, p.(Pro288Leu)	Hem	XLR	De novo	Pyruvate dehydrogenase E1-alpha deficiency (312170)	Novel	Pathogenic (PS2, PM1, PM2, PP2, PP3)
Renal abnormalities									
PKD2	9081/F	39	NM_000297.4:c.1837C>T, p.(Gln613*)	Het	AD	n/a	Polycystic kidney disease 2 (613095)	Known	Pathogenic (PVS1, PM2, PP3, PP5)
PKHD1	8084/M	2	NM_138694.3:c.8555-2A>G/ c.3907delG, p.(Val1303Serfs*13)	Comp Het	AR	Pat/Mat	Polycystic kidney disease 4, with or without hepatic disease (263200)	Known/ Novel	Pathogenic/Pathogenic (PVS1, PM2, PP3, PP4, PP5/PVS1, PM1, PM2, PM3, PP3, PP4)
Others (Immune abnormality, Germline tumor, Hematological abnormality, Endocrine abnormality, Sex abnormality)									
ARPC1B	8140/M	1	NM_005720.4:c.783G>A, p.(Ala261Ala)	Hom	AR	Mat/Pat	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease (617718)	Novel	Pathogenic (PVS1, PM2, PP1, PP3, PP4)
DICER1	9117/F	19	NM_030621.4:c.2685dupA, p.(Phe896Ilefs*5)	Het	AD	Mat (same phenotype)	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors (138800)	Novel	Pathogenic (PVS1, PM1, PM2)
RPS26	9071/M	2	NM_001029.5:c.181+1delG	Het	AD	n/a	Diamond-Blackfan anemia 10 (613309)	Novel	Pathogenic (PVS1, PM2, PP3)
THRA	8127/M	3	NM_003250.5:c.1416dupC, p.(Ser473Leufs*2)	Het	AD	n/a	Hypothyroidism, congenital, nongoitrous, 6 (614450)	Novel	Pathogenic (PVS1, PM2, PP3)

ZFPM2	9045/F (46,XY)	6	NM_012082.4:c.192T>G, p.(Cys64Trp)		Het	AD	Mat	46,XY sex reversal 9 (616067)	Novel	Likely Pathogenic (PM1, PM2, PP3, PP4)

Case ID: unique patient code
F: Female, M: Male
Het: Heterozygous, Comp Het: Compound Heterozygous, Hom: Homozygous, Hem: Hemizygous
AD: Autosomal Dominant, AR: Autosomal Recessive, XLD: X-linked Dominant, XLR: X-linked Recessive
Mat: Maternal, Pat: Paternal, n/a: not applicable

Table 2: Cases with a primary pathogenic variant consistent with their phenotype (Table 1) and a secondary VUS in a gene potentially associated with aspect(s) of the phenotype.

Case ID/ Gender	Age (yrs) at testing	Phenotype on referral	Primary finding (Table 1)	Secondary Gene	Secondary Variant	Zygosity	Inheritance mode	Segregation	MIM Disease (MIM number)	Novel/ Known	Classification (ACMG score)
8039/F	10	Sensorineural hearing loss and cardiomyopathy	***GJB2:c.35delG (Hom)***	***TGFB3***	NM_003239.4:c.785G>T, p.Gly262Val	Het	AD	n/a	Arrhythmogenic right ventricular dysplasia 1 (107970)	Novel	Variant of Uncertain Significance (PM2, PP3)
9081/F	39 (onset of symptoms 36y/o)	Renal abnormality	***PKD2:c.1837C>T (Het)***	***PKHD1***	NM_138694.3:c.6992T>A, p.Ile2331Lys	Het	AR	n/a	Polycystic kidney disease 2 (613095)	Known	Variant of Uncertain Significance (PM1, PM2, PP2, PP3, PP5)
9151/F	9	Delayed psychomotor development, poor speech, hypotonia, large ears, hypertelorism and	***SET:c.99_102delAGAC (Het)***	***MYH7***	NM_000257.4:c.5283+1G>A	Het	AD	n/a	Cardiomyopathy, dilated, 1S (613426)	Novel	Variant of Uncertain Significance (PVS1, PM2, PP3)

	left cardiac ventricular dilation

Case ID: unique patient code
F: Female, M: Male
Het: Heterozygous, Comp Het: Compound Heterozygous, Hom: Homozygous, Hem: Hemizygous
Mat: Maternal, Pat: Paternal, n/a: not applicable
DD: developmental delay