

Patient 1: final list of 11 candidate genes

Gene	Type	Variant
CENPF	Missense	c.G6136A:p.A2046T
UBA7	Missense	c.G2825A:p.R942K
SKIV2L	Missense	c.G1617A:p.M539I
TAP2	Missense	c.C1840T:p.R614C
EVX1	Missense	c.G41A:p.G14D
WIPF3	Missense	c.A1205G:p.Q402R
MBOAT4	Nonsense	c.C553T:p.R185X
SLC22A24	Missense	c.T1180A:p.C394S
SCYL2	Missense	c.A1940C:p.K647T
FBN1	Missense	c.C5183T:p.A1728V
C18orf54	Frameshift deletion	c.493delG:p.E165fs