Table e1: ACMG criteria for EBF3 variant classification

Patient	**Variant**	**rs ID (if available)**	**ACMG classification**	**ACMG criteria**
Patient 1	EBF3:c.512G>A (p.Gly171Asp)	rs1057519437	Pathogenic	PM2, PP2, PP3, PP5, PM1, PS2
Patient 2	EBF3:c.481delT (p.Cys161Alafs*21)	-	Pathogenic	PVS1, PM2, PS2
Patient 3	EBF3:c.512G>A (p.Gly171Asp)	rs1057519437	Pathogenic	PM1, PM2, PP2, PP3, PP5, PS2
Patient 4	EBF3:c.422A>G (p.Tyr141Cys)	rs1057519519	Pathogenic	PP5, PM2, PP2, PP3, PS2

Table e2: genes included in the 10q26 deletions

Patient	**Variant**	**rs ID (if available)**
Patient 5	arr[GRCh37] 10q26.2q26.3 (127657000_135434409)x1	FANK1, FANK1-AS1, ADAM12, RNA5SP328, SAR1AP, LINC00601, C10orf90, DOCK1, INSYN2A, NPS, FOXI2, BUB1P1, CLRN3, PTPRE, MKI67, LINC01163, LINC02667, MGM, LINC02666, EBF3, MIR4297, C10orf143, CTAGE7P, PPIAP3, GLRX3, LINC0264, MIR378, TCERG1L TCERG1L-AS1, LINC01164, PPP2R2D, BNIP3, JAKMIP3, JAKMIP3-AS1, DPYSL4, STK32C, LRRC27, PWWP2B, LINC02870, LINC01165, INPP5, NKX6-2, CFAP46, LINC01166, LINC0116, LINC0116, ADGRA1-AS1, ADGRA, RPL5P28, KNDC1, UTF1, VENTX, MIR202HG, MIR202, ADAM8, TUBGCP2, ZNF511, ZNF511-PRAP1, CALY, BANF1P, PRAP, FUO, ECHS, MIR3944, PAOX, MTG, SPRN, OR6L2, SCART1, OR7M1P, CYP2E1, SYCE1, OR6L1P
Patient 6	arr[GRCh37] 10q26.2q26.3 (128939462_135506781)x1	DOCK1, INSYN2A, NPS, FOXI2, BUB1P1, CLRN3, PTPRE, MKI67, LINC01163, LINC02667, MGM, LINC02666, EBF3, MIR4297, C10orf143, CTAGE7P, PPIAP3, GLRX3, LINC0264, MIR378, TCERG1L TCERG1L-AS1, LINC01164, PPP2R2D, BNIP3, JAKMIP3, JAKMIP3-AS1, DPYSL4, STK32C, LRRC27, PWWP2B, LINC02870, LINC01165, INPP5, NKX6-2, CFAP46, LINC01166, LINC0116, LINC0116, ADGRA1-AS1, ADGRA, RPL5P28, KNDC1, UTF1, VENTX, MIR202HG, MIR202, ADAM8, TUBGCP2, ZNF511, ZNF511-PRAP1, CALY, BANF1P, PRAP, FUO, ECHS, MIR3944, PAOX, MTG, SPRN, OR6L2, SCART1, OR7M1P, CYP2E1, SYCE1, OR6L1P, FRG2B, RARRES2P2, AGGF1P2, CLUHP5, DUX4L28, DUX4L25, DUX4L24, DUX4L23, DUX4L22, DUX4L21, DUX4L20 , DUX4L29, DUX4L10, DUX4L11, DUX4L12, DUX4L13, DUX4L14, DUX4L15