Appendix 6. Putative copy number variants identified in FECD cases that did not have an expanded repeat (A), had an expanded repeat (B) and in non-FECD controls (C).

HGNC gene symbol	Sample ID	Genomic interval (hg19)	CNV type	Transcript accession number	Number of exons	Reads expected	Reads observed	Reads ratio	BF
(A)									
SLC4A11	593	chr20:3208906-3211293	deletion	NM_032034.4	8	555	296	0.533	8.1
SLC4A11	831	chr20:3209485-3210906	deletion	NM_032034.4	5	83	37	0.446	6.72
(B)									
LOXHD1	968	chr18:44126859-44137335	deletion	NM_144612.7	2	307	192	0.625	5.34
(C)									
ZEB1	1147	chr10:31812862-31813041	deletion	NM_030751.6	1	36	7	0.194	5.33
AGBL1	1160	chr15:86814851-86814937	duplication	NM_152336.4	1	433	656	1.512	5.75
LOXHD1	1139	chr18:44126859-44127021	duplication	NM_144612.7	1	405	556	1.373	5.11
SLC4A11	1143	chr20:3214161-3214283	deletion	NM_032034.4	1	63	22	0.349	5.43

HGNC gene symbol, sample ID, genomic coordinates of variant in hg19 version of the genome, the type of copy number variant (CNV), transcript accession number, number of exons affected, the expected and observed reads and their ratio relative to each other, and the Bayesian factor (BF) score are shown. There was no evidence to support the CNVs when the samples were reviewed using the IGV software.