Table 2. Clinical features of individuals with ATP6V1B2 variants.

Individual/family	1 (n=3)	2 (n=3)	3 (n=2)	4 (n=1)[a]	5 (n=1)[a]	6 (n=1)[a]	7 (n=1)	8 (n=1)	9 (n=1)	10 (n=1)
***ATP6V1B2* Variant c.1516C>T**	+	+	+	+	+	+	+	+	+	+
DDOD	+	+	-	-	-	-	-	-	-	-
DOORS	-	-	+	+	+	+	+	+	+	+
Deafness	+	+	+	+	+	+	+	+	+	+
Onychodystrophy	+	+	+	+	+	+	+	+	+	+
Brachydactyly	+	+	+	+	+	+	-	+	+	+
Bulbous swelling of terminal phalanges	+	+	+	-	-	-	-	-	-	-
Finger-like thumb	+	+	-	-	-	-	+	-	-	+
Triphalangeal thumb	-	-	+	+	+	+	-	-	+	-
Dental anomalies	+[b] (2/3)	-	-	+[b]	+[c]	-	-	-	-	-
DD/ID	-	-	-	+[d]	+[e]	+[f]	+[d]	+[f]	+[e]	+[f]
Seizures	-	+ (1/3)	+ (1/2)	-	+	+	-	-	+	+

DD: developmental delay; ID: intellectual disability. [a]Individuals 4, 5 and 6 were reported by Campeau et al. and referred as individuals 24, 25 and 26, respectively in the previous publication; [b]Late dentition; [c]Misalignment; [d]Mild ID; [e]Severe ID; [f]Severity of ID not assessed. See supplement for other dysmorphisms noted.