

Supplementary Table 2: Comparison of clinical details of individuals with FBXO11 in our cohort vs clinical details of individuals with FBXO11 variants reported in the literature to date.

Paper	Martinez et al	Fritzen et al	Gregor et al	Jansen et al	Lee et al	Engelen et al	Gregor et al	Pan et al	sum	%
Year published	2017	2018	2018	2019	2020	2021	2022	2025		
Total No of cases	1	2	20	24	3	1	22	12	85	100.00
Number of new cases	1	2	20	22	3	1	22	12	83	97.65
Male	1	2	13	18		1		1	36	42.35
Female			7	6	3				16	18.82
No. of variants	1	2	20	24	1	1	22	5	76	89.41
missense		0	8	10			13		31	36.47
splice site		1	3	1				0	5	5.88
in-frame single aa deletion		0	0	2			1	1	4	4.71
partial/large gene deletions		0	2	2		1	9	3	17	20.00
nonsense		1	7	2	1				11	12.94
framshift	1	0	0	6					7	8.24
Clinical features									0	n=85
Short stature		1	4				3	6	14	16.47
macrocephaly					2		0		2	2.35
microcephaly	1	2	5		0		3		11	
Mild ID							15		15	
ID (borderline to severe ID)	1	2	20	24	3	1	22	12	85	100.00
Motor delay	1	2	15		2	1			21	24.71
speech delay	1	2	20		2	1		8	34	40.00
Behavioural	1	2	17	16	1	1	12	1	51	60.00
Autism	1	0	0	7					8	9.41
Anxiety		0	0	3					3	3.50

ADHD		2	0	5	1	1			9	10.59
aggression		0	0	3					3	3.50
seizures		0	5	3	1		10	3	22	25.88
Brain imaging		1	16	17	1				35	41.18
MRI brain abnormalities		1	5	6	1		8		21	24.71
Joint hyperlaxity		0	3	8					11	12.94
obesity		0	4	7			4	7	22	25.88
vision	1	1	6	9	1	1	10	4	33	38.82
sleeping		0	9						9	10.59
feeding difficulties		0	5	11					16	18.82
hypotonia		1	17	12			14	1	45	52.94
cleft lip		2	3					0	5	5.88
hand/feet anomalies	1	2	12	12				0	27	31.76
genital anomalies		2	1					0	3	3.53
Cardiac anomalies		0	4	2				0	6	7.06
Renal	1	0	1					0	2	2.35
Dysmorphism	1	2	20	24		1	21	0	69	
otitis media		0	4				9		13	
Hearing		0	0			0			0	