

Supplementary Table 3: Types of FBXO11 variants published in the literature thus far

Paper	Martinez et al	Fritzen	Gregor	Jansen	Lee	Engelen	Gregor	Pan	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
No. of variants	1	2	20	24	1	1	22	5	76	89.41
missense	0	0	8	10	0	0	13	0	31	36.47
splice site	0	1	3	1	0	0	0	0	5	5.88
in-frame single aa deletion	0	0	0	2	0	0	1	1	4	4.71
partial/large gene deletions	0	0	2	2	0	1	9	3	17	20.00
nonsense	0	1	7	2	1	0	0	0	11	12.94
framshift	1	0	0	6	0	0	0	0	7	8.24

