

Supplementary Material
for
PSEUDODIASTROPHIC DYSPLASIA EXPANDS THE KNOWN PHENOTYPIC SPECTRUM
OF DEFECTS IN PROTEOGLYCAN BIOSYNTHESIS

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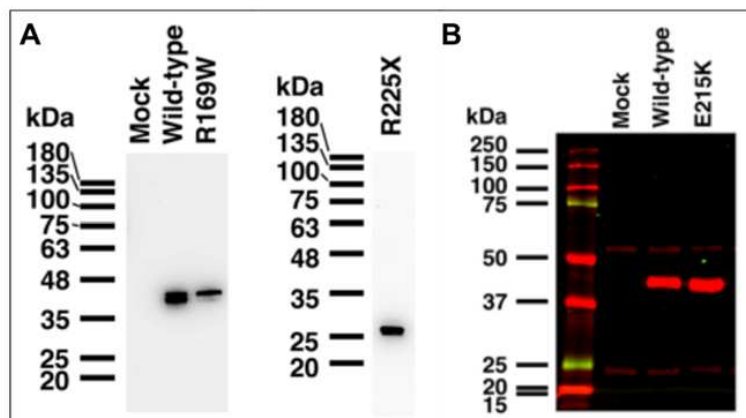


Figure S1. Western blot analysis of recombinant B3GAT3 and CANT1. (A) Expression level of recombinant B3GAT3 in HEK293T cell lysates. (B) Expression level of recombinant CANT1 in conditioned media from HEK293T cells.

SUPPLEMENTARY TABLE

Table S1. Coverage statistics for the exome sequencing results

Coverage statistics WES	1_Mother	1_Father	Patient 1A	Patient 1B	2_Mother	2_Father	Patient 2B
Average coverage (X)	109.9	84.9	65.8	69.7	147.6	148.3	124.2
% of targets covered >20X	89.1	88.6	83.2	85.3	94.0	93.9	93.5