

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

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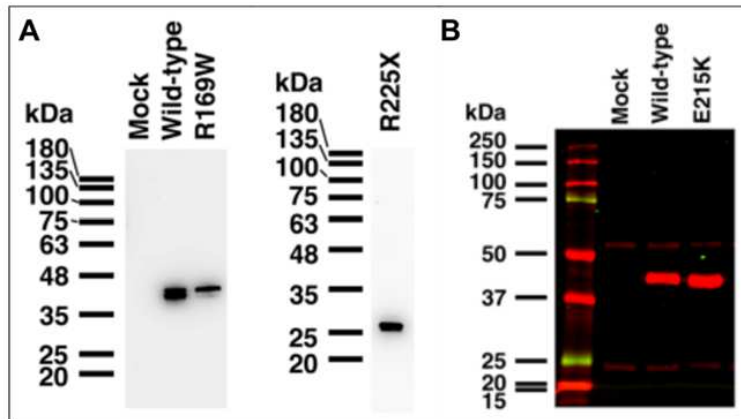
### 1. Supplementary Figure

- **Figure S1:** Western blot analysis of recombinant B3GAT3 and CANT1.

### 2. Supplementary Table

- **Table S1:** Coverage statistics for the exome sequencing results.

## SUPPLEMENTARY FIGURE



**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