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Supplemental Data

Biallelic Mutations in *NBAS* Cause Recurrent Acute Liver Failure with Onset in Infancy

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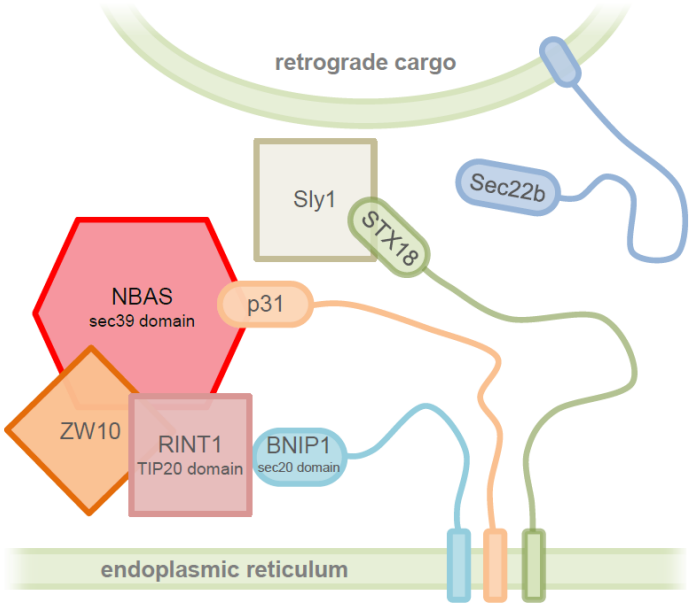

Figure S1. Proposed Function of NBAS
Schematic representation of the predicted NBAS function in the t-SNARE Syntaxin 18 complex.

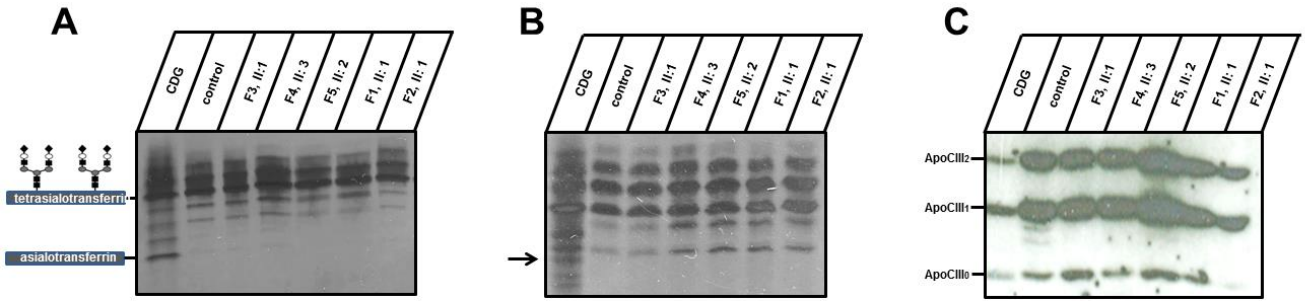

Figure S2. Isoelectric Focusing of Transferrin, Alpha-1-antitrypsin, and Apolipoprotein CIII
Sera of a control, a CDG patient and the individuals with *NBAS* variants were analyzed by isoelectric focusing (IEF), followed by in gel immunodetection.
(A) `Tetrasialo' and `asialo' indicate transferrin forms with four or no sialic acid residues on transferrin. A control-like IEF pattern was obtained for all individuals with *NBAS* variants. Filled square, N-acetylglucosamine; grey circle, mannose; open circle, galactose; filled diamond, sialic acid.
(B)For alpha-1-antitrypsin the position of the first additional abnormal cathodal band is indicated by an 'arrow'. This band and all bands below are abnormal and indicate a N-glycosylation deficiency in case of the CDG patient only.
(C) To further investigate, whether the variants in *NBAS* have an impact on the biosynthesis of core 1 mucin type O-glycans, IEF of Apolipoprotein CIII (ApoCIII) was performed. $ApoCIII_2$, $ApoCIII_1$ and $ApoCIII_0$ indicate the variability in the amount of sialic acid residues linked to ApoCIII. As for the N-glycosylation, no abnormalities were decteted in case of the individuals with *NBAS* variants.