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CASE REPORT

A Case of Fetal Familial Hemophagocytic Lymphohistiocytosis Type 5 caused by *STXBP2* Gene Mutation

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SUMMARY

Background: **Familial hemophagocytic lymphohistiocytosis type 5 (FHL-5) is a rare hyper-inflammatory syndrome caused by mutations in STXBP2. Most cases present at 2 - 6 months of age, and FHL-5 is extremely rare in neonates.**
Methods: **Appropriate laboratory tests, abdominal ultrasonography and whole exome sequencing were carried out. Respiratory support, antibiotics, and transfusion of blood products were done.**
Results: **Laboratory tests revealed metabolic acidosis, thrombocytopenia, mild anemia, and low fibrinogen level. Blood culture, metagenomics, and TORCH screening were negative. Liver and spleen enlargements were confirmed by abdominal ultrasonography. Whole exome sequencing identified a homozygous mutation in STXBP2 c. 1432del G (p. V478Sfs*5). The heterozygous STXBP2 mutation was identified in the paternal grandfather, maternal grandfather, and parents.**
Conclusions: **Here we report a case with a novel homozygous deletion in exon 16 of *STXBP2*, which caused the earliest reported case of FHL-5 in a neonate. Our results identify a new pathogenic variant for the early identification and clinical consultation of FHL-5.**
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Supplementary Data

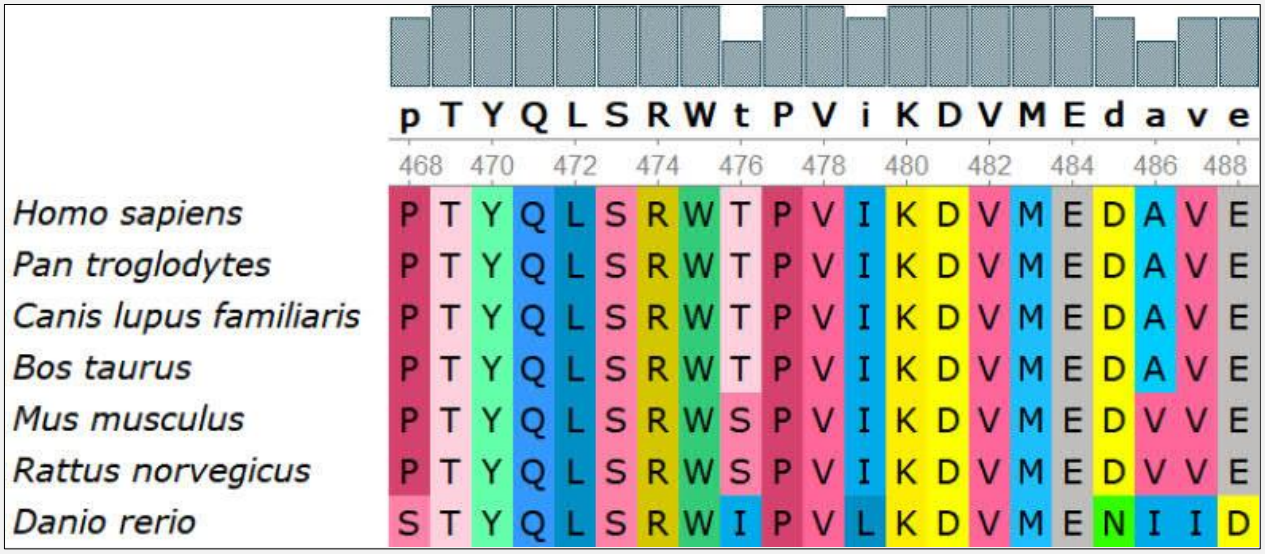

Figure S1. Protein alignment of *STXBP2* in various species. Val 478 is highly conserved in the species examined.

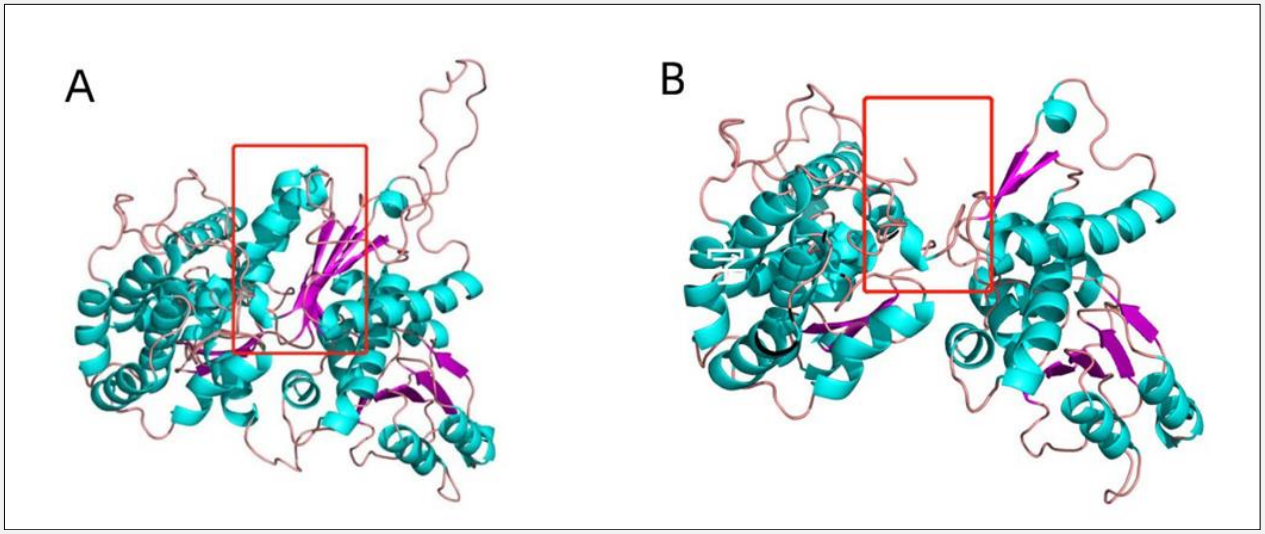

Figure S2. Predicted structure of STXBP2 protein.

(A) STXBP2 wild-type protein and (B) STXBP2 mutant protein.