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## **Bioinformatic prediction revealed the potential effects of *THBS1* mutation on liver fibrosis and inflammation**

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### **Abstract:**

Biliary atresia (BA) is a rare, life-threatening inflammatory disease of the liver and bile ducts that occurs in newborns. Patients with BA may develop profound liver fibrosis, leading to liver transplantation. We performed whole genome sequencing on the patients with BA and their unaffected parents. Bioinformatic analysis, including protein-protein interaction network and hub genes analysis, identified *THBS1* (Thrombospondin 1) as the most vital hub gene of the candidate disease-causing genes of BA. Here, we identified a compound heterozygous variant in one BA trio. Protein structure prediction using AlphaFold and SWISS-MODEL suggested that the compound heterozygous variant on *THBS1* could affect the tertiary structure, stability, or calcium ions environment of THBS1. From the RNA-seq data, we observed differential expression between BA patients and normal controls of *THBS1* in the human liver (Luo et al. 2019). Moreover, we also observed abnormal expression of *THBS1* in the murine model of biliary atresia from a previously published paper (Bessho et al. 2014). To investigate the potential mechanism of cell activities on BA, we applied the gene deconvolution method to impute cell type-specific expression from bulk RNA-seq data of BA and normal controls. Notably, *THBS1* showed dysregulation in BA samples in hepatocytes and inflammatory macrophages. Together with a literature review on *THBS1*, we hypothesized that dysregulation of *THBS1* and its associated pathways would promote liver fibrosis and inflammation, contributing to the etiology and progression of BA.

Author Disclosure Information:

**Q. Lin:** None. **C.S. Tang:** None. **P.H. Chung:** None. **V.C. Lui:** None. **P.K. Tam:** None.

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