

Supplementary file 1. Example of patient presentation

Initial Presentation:

Basic information: A 13-year-old boy

Clinical presentation: Recurrent fever for more than 1 month. The first fever occurred without obvious causes in early September 2021, and the maximum temperature reached 39.0°C accompanied by headache. He was treated with anti-infective treatment in the community clinic for 4 days. Then the temperature returned to the normal range. Two weeks later, the patient got a fever above 39.0°C again, and the intervals was gradually shortened. The temperature returned to normal after 5 days of treatment with ceftriaxone and methylprednisolone for septicemia. However, no further examination was made to determine the cause of fever. The night before coming to our hospital, the boy had a higher temperature once again. During the course of illness, he had poor appetite and sleep. The temperature rose to as high as 40.3°C on the 2nd day. Empirical anti-infective treatment with cefotaxime (1.0 g q6h) was applied to the patient. 48 h later, observation on the temperature showed a constant fever over 40.0°C. Meanwhile, the tenderness in the right upper abdomen worsened.

Physical Examination: A physical examination was remarkable for mild tenderness in the upper abdomen, but no rebound tenderness. Besides, the liver and spleen were palpable 4.0 cm under the ribs.

Relevant medical history: He was born in a non-consanguineous family.

Initial lab results: The levels of inflammatory biomarkers showed just a slight increase, while the coagulation, liver function and tumor markers were in the normal rang.

Complete Presentation

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Additional Diagnostic Test: Abdominal ultrasound showed multiple cysts in the liver and hepatosplenomegaly, no obvious renal cyst was found. Abdominal computerized tomography (CT) scan was performed, which showed prominent dilation of the intrahepatic bile ducts. To make sure of the diagnosis, a magnetic resonance cholangiopancreatography (MRCP) was performed, which revealed multifocal dilatation of the intrahepatic bile ducts associated with hepatomegaly. Both CT and MRCP showed obvious "central spot sign." Based on these imaging findings, a diagnosis was considered. Genetic testing for the child and his parents were not performed.