A rare case of hereditary thrombotic thrombocytopenic purpura in a toddler

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Background:

Hereditary thrombotic thrombocytopenic purpura (hTTP) is a thrombotic microangiopathy caused by pathogenic variants in the ADAMTS13 gene resulting in highly reduced activity of von Willebrand factor-cleaving metalloprotease. hTTP is characterized by systemic formation of platelet-rich thrombi in microvasculature resulting in organ ischemia, thrombocytopenia, and microangiopathic hemolytic anemia.

Case presentation:

A 21-month-old female toddler was hospitalized because of thrombocytopenia (38/μL) during acute respiratory infection. On physical examination, the patient was subfebrile with a few isolated petechiae on the face and thighs. In her history, she had transient neonatal thrombocytopenia that spontaneously resolved, and the workup excluded autoimmune etiology. She wasn't regularly vaccinated because of her parent's decision. Later, she also developed hemolytic anemia (hemoglobin 85 g/L, LDH 756 IU, reticulocytes 4.2%) while the blood smear showed schistocytes. Urinalysis showed proteinuria and microhematuria. A platelet transfusion was administered. Further workup excluded immune thrombocytopenia and revealed very low ADAMTS13 activity (<1%) that is consistent with a diagnosis of TTP. Plasmapheresis was performed on two consecutive days with prompt elevation of platelet level. Meanwhile, next-generation gene sequencing from a peripheral blood sample found two missense heterozygotic variants in the ADAMTS13 gene confirming hTTP diagnosis. Prophylactic fresh frozen plasma (FFP) was continued every three weeks. During follow-up, she had one relapse during the flu and was successfully treated with FFP. A special program of vaccination is recommended.

Conclusion:

hTTP is an extremely rare (0.5-2/10^6) hematologic disorder primarily seen in neonates and children. It is characterized by exacerbations following acute infections. Therefore, it is of great importance to observe platelet dynamics and administer FFP if needed during any infective episodes to prevent potentially fatal consequences of end-organ dysfunction.

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