

Prothrombin Gene 20210A mutation heterozygosity and MTHFR Gene C677T mutation homozygosity detected in a male toddler experiencing femoral venous thrombosis during diabetic ketoacidosis



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Introduction:

Diabetic ketoacidosis (DKA) as an inflammatory state combined with the disruption of the normal coagulation cascade can lead patients to an increased risk of thrombosis. Especially, patients that are genetically susceptible to thrombosis could develop deep venous thrombosis (DVT) due to inflammation, dehydration, and hyperviscosity secondarily to DKA. It is noteworthy that children with DKA who underwent central venous catheter placement could develop DVT, especially those that are less than 3 years old. This can be explained due to smaller vessel diameter and worse presentation of the illness at the beginning.

Purpose:

To describe femoral venous thrombosis during the course of severe ketoacidosis in a male toddler diagnosed with T1D.

Case report:

A 26/12 years old boy was admitted to our hospital with severe DKA (pH 6.95, HCO₃ 4mmol/L) and hyperglycemia (485mg/dl) as well as extremely dehydrated. Despite the applying of current guidelines for the management of DKA, the patient was deteriorated and transferred to PICU. In the PICU he was intubated and a central venous catheter was inserted in the right femoral vein. On the 4th day of hospitalization a swelling on his right thigh developed accompanied by fever. A femoral venous thrombosis was confirmed by a Doppler ultrasound and a medication with low molecular weight heparin (LMWH) administrated. Despite the heparin administration the anticoagulant response (anti-factor Xa activity) was poor. A further investigation concerning thrombophilia screening was performed, which revealed a prothrombin Gene 20210A mutation heterozygosity and MTHFR Gene C677T mutation homozygosity. Subsequently the dose of LMWH was increased and 10 days later the levels of anti-Xa activity were normalized. One month later the child has a good glycemic control and a normal anti-factor Xa activity. All family members are recommended to be screened for thrombophilia.

Femoral venous thrombosis is a rare complication of DKA in children, associated with the use of central venous catheter; however, in our case an undiagnosed thrombophilia further predisposed to coagulopathy. Although the above findings are rare, a high index of clinical suspicion for thrombotic episodes is required in severe forms of DKA. Thrombophilia screening should be considered in selected cases.

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