Lower Limb Oedema in Beckwith-Wiedemann Syndrome

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Abstract : We present a case of inferior vena cava agenesis (IVCA) associated with bilateral deep venous thrombosis (DVT) in a patient with Beckwith-Wiedemann syndrome (BWS). In adult patients with BWS presenting with bilateral lower limb oedema, specific aetiological factors should be considered. These include cardiomyopathy and intraabdominal tumours. Congenital malformations of the IVC, through causing relative venous stasis, can lead to lower limb oedema either directly or indirectly by favouring lower limb venous thromboembolism; however, they are yet to be reported as an associated feature of BWS. Given its life-threatening potential, the prompt initiation of treatment for bilateral DVT is paramount. In BWS patients, however, this can prove more complicated. Due to overgrowth, the above-average birth weight can continue throughout childhood. In this case, the patient's weight reached 170 kg, impacting on anticoagulation choice, as direct oral anticoagulants have a limited evidence base in patients with a body mass above 120 kg. Furthermore, the presence of IVCA leads to a long-term increased venous thrombosis risk. Therefore, patients with IVCA and bilateral DVT warrant specialist consideration and may benefit from multidisciplinary team management, with hematology and vascular surgery input. Conclusion: Here, we showcased a rare cause for bilateral lower limb oedema, respectively bilateral deep venous thrombosis complicating IVCA in a patient with Beckwith-Wiedemann syndrome. The importance of this case lies in its novelty, as the association between IVC agenesis and BWS has not yet been described. Furthermore, the treatment of DVT in such situations requires special consideration, taking into account the patient's weight and the presence of a significant, predisposing vascular abnormality.

Keywords : Beckwith-Wiedemann syndrome, bilateral deep venous thrombosis, inferior vena cava agenesis, venous thromboembolism

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