APNEIC EVENT IN WOLF-HIRSCHHORN SYNDROME: COULD THIS HAVE BEEN A 'TET' SPELL?

Kene Maduemem*, Gheetha Arumugam, Amol Chingale. Lincoln County Hospital, Lincoln, UK

10.1136/archdischild-2019-epa.439

Background Wolf-Hirschhorn syndrome (WHS) is a rare genetic condition caused by partial deletion of short arm of chromosome 4. Clinical manifestations comprise but not limited to typical facie, mental retardation, growth delay, epilepsy, and congenital cardiac defects. Congenital heart defects occur in about 25–50% of cases. We describe a child with WHS presenting with a femoral fracture; he became apnoeic in the course of management resulting in a cascade of events.

Case Report A 4.5-year-old boy known to have WHS was admitted with left femoral fracture. The syndrome was an antenatal diagnosis. Other associations noted were severe growth delay (weight of 6 kg), epilepsy, global developmental delay, visual impairment, and tetralogy of Fallot (TOF). His TOF has been managed uneventfully on propranolol with acceptable oxygen saturation (Sp02) in the 70s. The spontaneous fracture required Gallows traction, which provoked distress. He developed hypoxic crisis requiring oxygen therapy to maintain acceptable SpO2. Subsequently, he went apnoeic and dusky needing bag and mask ventilation. Blood gas revealed severe metabolic acidosis, which improved after fluid bolus. Morphine dosing regimen for pain control was continued. Knee-chest position could not be attempted due to the fracture. Although he got treated for suspected sepsis due to the dilemma of events, he did have a hypercyanotic spell in hindsight. He was subsequently discharged with conservative management of the fracture.

Conclusion To the best of our knowledge, this is the first reported case of hypercyanotic (Tet) spell in WHS. This rather uncommon event resulted in a significant learning point for medical and nursing staff.

21 MONTH OLD INFANT WITH SEVERE HYPONATREMIA CAUSED BY EXCESSIVE WATER INTAKE DURING FEBRILE INFECTION. MANAGEMENT AND DISCUSSION OF THE CASE

1Anastasia Anastasiou-Katsiarianni*, 1Konstantinos Tsakos, 1Angeliki Besika, 1Maria Lampri, 1Kalliopi Tanou, 2Michail Kastamoulos, 2Evaggelia Lykopoulou. 1Paediatric Ward, 2Asklepieio General Hospital of Volos, County of Magnesia, Volos, Greece; 1First Department of Paediatrics, ‘Agia Sofia Children’s Hospital’, Athens, Greece

10.1136/archdischild-2019-epa.440

Introduction Hyponatremia is the most common electrolyte disorder, defined as a serum sodium level less than 135 meq/L. It is classified according to serum sodium concentration as mild, moderate and severe and according to its manifestation time in acute (<48 hours) and chronic. It can lead to a wide range of symptoms, from mild to severe, even life-threatening (cerebral edema).

Purpose Discussion and management of a case of a 21 month old female infant with severe hyponatremia (Na: 117 meq/L), presented with drowsiness accompanied by polydipsia and polyuria following a febrile infection.

Clinical case The infant presented with drowsiness started 12 hours ago, accompanied by polydipsia (water intake >7 Lt/day), polyuria and high temperature up to 39.8 °C that started 3 days ago. From the medical history high water intake is reported from the age of 6 months old. At the time of admission, she was haemodynamically stable, afebrile, presented with tonsillitis and signs of mild dehydration. Laboratory tests revealed severe hyponatremia (Na: 117 meq/L), dehydration with normal renal function tests, low serum and urine osmolality and normal white blood cell count and CRP. Kidney-ureter-urinary bladder-adrenal gland ultrasound and fundoscopy showed no pathological findings. The patient was treated with intravenous fluids based on the hypotonic dehydration treatment protocol, with continuous neurological, blood gas and biochemical monitoring. Serum sodium levels recovered to normal (Na: 136 meq/L) over the following 24 hours. On the third day of hospitalization she was transferred to the Childrens’ Hospital of Athens ‘Agia Sofia’ in order to complete further investigations to exclude possible adrenal or pituitary gland dysfunction and cystic fibrosis disease, which revealed no pathological findings. As a result, the most likely cause of hyponatremia was considered to be the excessive water intake.

Conclusions We presume high water intake as the most likely cause of hyponatremia for our patient, after having excluded other causes of hyponatremia. Although high fluid intake is considered to be a rare cause of hyponatremia in childhood, based on published data, it has to be included in the differential diagnosis. The detailed medical history, the clinical examination and laboratory tests lead to early diagnosis. The cautious correction of serum sodium levels, as well as of co-existent acid-based disorders, is the mainstay of successful treatment of such a potentially life-threatening condition.