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

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 chest position could not be attempted due to 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.

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), 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 Children’s 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 co-existing acid-based disorders, is the mainstay of successful treatment of such a potentially life-threatening condition.