GP60

CLINICAL UTILITY OF ELEVATED VITAMIN B12 LEVELS WITH A PROGRESSIVELY ABNORMAL BLOOD FILM

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10.1136/archdischild-2019-epa.126

Introduction Cobalamin plays an essential role in haematopoiesis, cell metabolism, production of DNA and neuronal function. High serum concentration of cobalamin is a common but underestimated finding. The aetiology of hypercobalaminaemia includes excess vitamin B12 intake, solid neoplasms, haematological malignancies, liver disease, renal failure and autoimmune diseases. Paradoxically, hypercobalaminaemia can be associated with a functional B12 deficiency. A finding of high serum cobalamin should prompt an early and in-depth search for these entities to ensure a favourable prognosis.

Case presentation We present a case of a thirteen-year old girl who presented with non-specific symptoms and an incidental finding of a raised serum cobalamin (1400 pg/mL) with initially only minor full blood count (FBC) abnormalities. Upon her assessment in the haematology clinic in OLCHC, repeat FBC results showed progressive anaemia with macrocytosis, a falling platelet count and serum cobalamin concentration of >6000 pg/mL. On repeat the cobalamin had increased to 10926 pg/mL. Urinary methylmalonic acid, plasma total homocysteine, liver and renal function were unremarkable, and she was not taking supplements or oestrogens. A bone marrow aspirate showed findings consistent with early diagnosis of Acute Myeloid Leukaemia (AML).

Conclusion This case highlights the importance of hypercobalaminaemia which should be followed by the search for the cause of this finding as early diagnosis can be an important prognostic factor. A possible malignant blood disorder should be considered when serum cobalamin concentrations are above the reference range and where increases due to supplements, inflammatory, renal or liver disease have been excluded. This underscores the importance of laboratories offering numerical values (rather than reporting high results as greater than a cut-off value) for elevated vitamin B12 levels in cases with minor but progressive FBC changes. An increase in the level of B12 may indicate the need for prompt referral to a haematologist for assessment and monitoring.

GP61

SWOLLEN EYES WITH AN UNPLEASANT SURPRISE – A CASE REPORT OF A STEROID RESISTANT NEPHROTIC SYNDROME

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10.1136/archdischild-2019-epa.127

With a reported incidence of 7:100 000, nephrotic syndrome (NS) is one of the more common nephrology disorders that can present to and is often treated by the general paediatrician in a primary and secondary level hospital. Standard treatment consists of steroids. Steroid resistant NS (SRNS) is an indication for renal biopsy.

We present a case of a 5-year-old girl, who presented to a regional hospital at the age of 2.5 years, with periorbital oedema and abdominal swelling. Her medical history was

significant for premature delivery, history of Intrauterine Growth Restriction (IUGR) and postnatal failure to thrive, persistent leucopenia and mild developmental delay. Upon presentation she had marked proteinuria, serum hypoalbuminaemia and hyperlipidaemia. A diagnosis of NS was made but she failed to respond to standard steroid treatment. She was transferred to a tertiary hospital on day 26 of admission with a suspicion of thrombophlebitis of her left lower limb. There, she was noted to have low set ears, areas of hyperpigmentation on her trunk, periorbital and limb oedema, ascites and abnormal posture. Pelvic X-ray showed bilateral hip dysplasia. Eye exam revealed retinal degeneration and blood investigations noted hypogammaglobulinemia, low CD4 and CD8 lymphocytes and high TSH. In view of her clinical picture, steroid resistant NS and leucopenia, Shimke Immuno-Osseous dysplasia (SIOD) was suspected. SIOD is a rare autosomal recessive disease consisting of renal failure, skeletal dysplasia and T-cell deficiency. This was subsequently confirmed by genetic testing which showed 2 pathogenic mutations within the SMARCAL 1 gene. Overtime she progressed to end stage renal disease, and initiated peritoneal dialysis within a year of diagnosis. She remains to date without any major infectious, central nervous system (CNS) or autoimmune complications commonly seen in SIOD.

The majority of paediatric patients with NS respond to steroid treatment. A subset of children might however have clinical and biochemical features or a family history predictive of a steroid resistant NS. Recent advances in molecular genetics identified underlying genetic cause in a large proportion of patients with SRNS.

These children might benefit from an early referral to a tertiary centre in order to accelerate the diagnosis, potentially avoid renal biopsy and limit the exposure to steroids, which won't improve their condition. Rather they might increase their already elevated risk of infection and other complications.

GP62

LIGNOCAINE TOXICITY: A CASE REPORT OF ADVERSE EFFECT OF LOCAL ANAESTHESIA IN COMMUNITY SETTING

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10.1136/archdischild-2019-epa.128

Introduction Lignocaine is an amide-type local anaesthetic and a class Ib antidysrhythmic agent, available since 1948. Systemic exposure to large amounts leads to adverse effects on the cardiovascular and central nervous systems (CNS). Local anaesthetics are extremely useful for providing anaesthesia and analgesia for paediatric population of all ages. Our aim is to raise awareness among the clinicians regarding the safety of administration of local anaesthetics.

Case presentation 11 weeks old baby boy brought into the Paediatric Assessment Unit (PAU) by ambulance following an episode of respiratory depression and generalised tonic clonic seizures in General Practice (GP) surgery. The patient had undergone a circumcision under local anaesthesia (brand name Xylocaine 1%) shortly prior to the event. He required basic life support to maintain airway and breathing prior to ambulance arrival. Two doses on intranasal midazolam were administered by the paramedics to abort the seizures. A Guedel airway was inserted for airway protection and 100% oxygen was supplied through the non-rebreather mask.