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Chemical Pathology

AN INTERESTING CASE IN CLINICAL BIOCHEMISTRY

Gemma M. Daley, David M. Kanowski, Lee A. Price
Department of Biochemistry, Sullivan Nicolaides Pathology, Brisbane, Qld, Australia

A 38-year-old male presented with a constellation of vague symptoms including memory loss. Our records demonstrated a history of worsening hyperphosphataemia over the preceding 3 years.
Of note were the pituitary function test results. The patient demonstrated hypogonadotrophic hypogonadism, elevated free thyroid hormones with an inappropriately normal thyroid stimulating hormone (TSH) concentration and a markedly elevated IGF-1 consistent with acromegaly. Their prolactin concentration was within the reference interval.
One of the challenges resulting from the current pandemic is the increase in phone consults which do not allow for a thorough clinical assessment. The referrer had not been able to review this patient in person due to their location being in a strict lockdown for much of the preceding 2 years.
This presentation follows the investigation and diagnosis of an interesting case in clinical biochemistry.

A CASE OF POST-COVID VACCINATION HYPONATRAEMIA

Andra Desra, Joel Smith, Cherie Chiang
Department of Pathology, Royal Melbourne Hospital, Parkville, Vic, Australia

Background: Syndrome of inappropriate anti-diuresis hormone (SIADH) can arise from pain, malignancy, medication use and infection including vaccines and COVID-19 infection. We describe a case of SIADH characterised by a severe acute hyponatraemia with confusion following a 2nd dose of COVID-19 mRNA vaccine.
Case presentation: 24 hours post-vaccination, a previously well 48-year-old female presented to the Emergency Department with sudden-onset acute confusion, dizziness and headache. On examination, she had right lateral gaze and was disoriented to time and place with normal vital signs. No abnormalities were detected on radiological investigation. Laboratory investigations on admission revealed a serum sodium of 113 mmol/L, glucose 7.3 mmol/L, serum copeptin 4.4 pmol/L. Post IV saline, she had rapid diuresis and rise of serum sodium to 127 mmol/L within 4 hours of admission; urine collected then revealed urine osmolality 81 mOsm/kg and urine sodium <20 mmol/L. CSF chemistry revealed a mildly elevated total protein. Cortisol, TSH, blood cultures, viral respiratory PCR, and CSF cultures were negative. The patient was not on any regular medications including diuretics.
Hyponatraemia has not been reported as a common side effect with COVID-19 vaccine. Following extensive investigation for other causes of hyponatraemia, we report a case of vaccine-related SIADH.

X-LINKED ADRENOLEUKODYSTROPHY: A BIOCHEMICAL CAUSE OF DEVELOPMENTAL REGRESSION IN A SEEMINGLY WELL CHILD

R. Campbell Heron, Campbell Kyle
Department of Chemical Pathology, Auckland City Hospital, Auckland, New Zealand

This case report outlines the diagnostic journey of a child with X-linked adrenoleukodystrophy (X-ALD) and discusses the prevalence, investigation and treatment of the condition. X-ALD is a rare condition, with a combined prevalence of heterozygotes and hemizygotes of about 1 in 17,000 children.[1] It causes a peroxisomal fatty acid oxidation disorder that leads to inflammatory disease in the cerebral white matter. Patients may present with developmental arrest or regression and language difficulties, though this may be misidentified as an attention deficit disorder. Childhood manifestations of the disease are irreversible and may be insidious. Medical intervention may help to slow or halt disease progression, making rapid recognition and treatment key to a favourable outcome.

Reference

1. Bezman L, Moser AB, Raymond GV, *et al.* Adrenoleukodystrophy: incidence, new mutation rate, and results of extended family screening. *Ann Neurol* 2001; 49: 512–7.

REDUC3 THE FAT – BEYOND GENE THERAPY

Myron Lee
Department of Chemical Pathology, Royal Prince Alfred Hospital, Sydney, NSW, Australia

The case is of a young gentleman with a genetic cause of severe hypertriglyceridaemia, resulting in recurrent pancreatitis. Conventional treatment was unable to bring his triglycerides down to safe levels. The recent emergence of RNA therapies such as short interfering RNA targeting apolipoprotein C3 show promise in phase 1 studies. A thorough understanding of the pathways of lipid metabolism is essential in understanding current and novel treatments as well as in the development of potential future therapeutics.

A TALE OF TWO SISTERS

Ranita Siru
Department of Biochemistry, Western Diagnostic Pathology, Jandakot, WA, Australia

A 15-year-old female was referred for gynaecology review for primary amenorrhoea. She had otherwise had a normal developmental trajectory through childhood. Family history was remarkable for primary amenorrhoea in her 17-year-old sister, as well as infertility in several maternal aunts. On examination, the patient was phenotypically female, and had Tanner stage 4 breast

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