GP53

ELUSIVE CAUSE FOR A RECURRENT FEVER IN A 2-YEAR-OLD BOY

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Introduction Periodic fever syndromes (PFSs) can present with a myriad of nonspecific signs and symptoms, with acute onset fever as the hallmark.

Case description We present the case of a 2-year-old boy who was admitted on 3 occasions with episodes of sudden onset isolated fever (40°C). From his history we noted 2 more hospital admittances in another medical center with similar presentations; also, the mother reported 4 more isolated fever episodes, the first at 4 months of age, during which the fever was apparently uninfluenced by antipyretics and disappeared after 3–5 days (no inflammatory markers were taken then as he was treated as an outpatient). A detailed history showed a clear periodicity of the episodes, occurring at 28–30 days intervals. Other symptoms were intermittently present during episodes: macular rash, unilateral cervical adenopathy (2 cm). Fever duration was 3 to 5 days, without a specific response to antipyretic or antibiotic treatment. A slight response to oral prednisone therapy was noted. At each admission, preliminary laboratory studies showed a marked inflammatory response (high CRP, procalcitonin, VSH, WBC). Broad spectrum antibiotic treatment was promptly started after blood and urine were sampled for cultures, as the presentations were interpreted at that time as high suspicion of occult sepsis/bacteremia. All sources of infection were ruled out each time by negative cultures. Blood smears and imaging studies did not raise any suspicion of malignancy. Tests for a primary immunologic defect were negative. PFSs were partially excluded as ANA titers, rheumatoid factor, IgD level was normal (HIDS), urinary mevalonic acid (MKD) was undetectable. Also, a genetic panel including 32 gene mutations/variants associated with PFSs including Familial Mediterranean Fever, CAPS, TRAPS, Blau syndrome, HIDS, ELANE related neutropenia and PAPA, did not detect mutations. All inflammatory markers returned to normal in between fever episodes. Our patient developed 3 more similar episodes, then the symptoms spontaneously stopped. The child is presently healthy, 6 months apart from the last episode.

Discussion PFSs are a diagnostic challenge. The key to a correct clinical approach is close monitoring of episodes and extensive workup. It seems that some causes remain undetermined despite diagnostic efforts or, as in our case, have a self-limited evolution. Future research in this pathology is still necessary.

GP54

SLEEPING SICKNESS: CONGENITAL CASE ASSOCIATED WITH A POSSIBLE SEXUAL TRANSMISSION

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Human African trypanosomiasis (HAT), also known as sleeping sickness, is caused by protozoan parasites transmitted by

the bite of a tsetse fly. It is characterized by an early stage, during which trypanosomes circulate in the blood or lymphatics, and a late stage, in which there is involvement of the central nervous system. In a globalised world, some cases are also diagnosed outside endemic African countries, thus HAT should be considered in differential diagnosis for travellers, tourists and migrants.

The authors report a rare case of a 19-year-old Afro-Brazilian boy, born in the USA, who was diagnosed with a congenital infection by *Trypanosoma brucei gambiense* at 16 months of age after returning to Portugal. Neither the patient nor his mother had ever been to Africa. He was admitted in our Paediatric Department for investigation of a wasting syndrome, lymphadenopathies and intermittent fever. An initial evaluation showed anaemia, hypergammaglobulinemia and a positive serology for cytomegalovirus infection. A month later the wasting syndrome had become worse and daytime somnolence was identified. At this point, the diagnosis of HAT was established in his mother, motivating specific diagnostic procedures in the child. Trypanosomes were detected in his blood and a CATT (Card Agglutination Test for Trypanosomes) was positive. A lumbar puncture established the diagnosis of late stage HAT. *Electroencephalogram* and brain MRI showed extensive brain damage. Fundoscopic examination revealed spots of an atypical chorioretinitis, possibly in association with the combined trypanosomal and cytomegalovirus infection. Treatment was established with intravenous eflornithine (DFMO). The drug was well tolerated, resulting in a gradual clinical improvement, including the complete regression of the brain and retinal changes. Two years after DFMO administration parasitological cure was confirmed according to established criteria. At the moment, the patient remains well, and show a normal psychomotor development.

The absence of suggestive epidemiological data in our patient and his mother made it difficult to establish the correct diagnosis. The correct diagnosis of our patient's condition was possible only after the etiology of his mother's disease was correctly established in our hospital. Given the available epidemiological data, transmission of HAT between our patient's parents must have occurred during sexual intercourse and is the first description of a case of probable sexual transmission of the disease. In our case, since neither the mother nor her son had ever been to Africa, the congenital transmission of HAT is indisputable.

GP55

GOING OUT ON A (CLOTTED) LIMB – AN INTERESTING CASE OF PAGET-SCHROETTER SYNDROME IN A YOUNG ADOLESCENT

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Introduction A 13-year-old female, presented to the Paediatric Emergency Department (PED), with a 4 week history of bruising to her right upper limb. There was no concurrent history of trauma to the limb. Bruising was progressive, and the arm had become increasingly painful in the three days prior to

presentation. On the night prior to presentation, swelling of the arm was noted with associated pain, which was worse on movement. Paraesthesia of the fingers of the affected limb was also noted.

Past history Her past history was notable for asthma (mild, no previous admissions) and hypermobility syndrome.

Medications There was no known drug allergies. She was not on any regular medications, including any form of contraceptive.

Vaccinations were up to date. Family history was notable for Raynaud's disease. There was no history of venous thromboembolism or any coagulation disorders.

Examination On examination, a visibly swollen right upper limb was noted, with a 3 cm difference in circumference and bruising as described. Dilated tortuous veins were visible. The patient was tender to palpation over the biceps muscle and site of tendon insertion. Full power and range of motion was noted at both shoulder and elbow joints. Radial pulses were present and equal. The remaining systemic examination was unremarkable.

Investigations Bloods normal, including coagulation

US Doppler upper limb no obvious DVT. However, dilatation of basilic vein proximally, with a possible varicosity was noted

MRA right upper limb large thrombus of right subclavian vein, protruding into right brachiocephalic vein and axillary vein

CT pulmonary angiogram: significant reduction in costoclavicular distance (right>left), suggestive of a thoracic outlet obstruction

Treatment Therapeutic subcutaneous low molecular weight heparin was commenced. The patient was admitted for further management, which included thrombectomy and heparin infusion.

Discussion Paget-Schroetter syndrome, or primary 'spontaneous' DVT of the upper limb, is a condition which typically affects otherwise healthy individuals. It occurs as a result of anatomical abnormalities of the thoracic outlet, which results in compression of the axillosubclavian venous system and subsequent thrombosis. It is a rare condition that is, however, important to recognize promptly as early detection and treatment have been found to reduce long-term sequelae.

This case, while rare, highlights the importance to us, as paediatric physicians, to consider in differential diagnoses, disorders and problems that would typically be more attributable to adult populations.

GP56

SEVERE HYPERCALCEMIC CRISIS IN AN INFANT WITH IDIOPATHIC INFANTILE HYPERCALCEMIA CAUSED BY MUTATION IN CYP24A1 GENE

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Introduction The presence of CYP24A1 mutations explains the increased sensitivity to vitamin D in patients with idiopathic infantile hypercalcemia and is a genetic risk factor for the development of symptomatic hypercalcemia that may be

triggered by vitamin D prophylaxis in otherwise apparently healthy infants.

Case report We present a case of a 4-month old girl who was initially hospitalized for severe hypotonia, lethargy and failure to thrive. The patient's history revealed recurrent vomiting. On the clinical examination we also noted a high forehead, a high arched palate and short metacarpal bones. Blood tests showed hypercalcemia, low PTH and phosphate and high vitamin D levels. Renal ultrasound showed medullary nephrocalcinosis. Correlating the clinical examination with the blood work vitamin D intoxication, hyperparathyroidism and Jansen's metaphyseal dysplasia were considered and ruled out. Genetic testing was performed and a compound heterozygote state was identified (mutations p.E143del and p.R396W) confirming the diagnosis of idiopathic infantile hypercalcemia. Both mutations have been formerly identified as loss-of-function mutations in the vitamin D-24-hydroxylase gene. Rehydration and furosemide therapy was applied and resulted in the normalization of calcium values and clinical improvement. On the long term the patient followed a low calcium diet and vitamin D supplementation was discontinued.

Conclusion Clinical symptoms, such as failure to thrive, vomiting, increased thirst, anorexia, hypotonia should always bring in discussion the possibility of hypercalcemia in order to diagnose and treat idiopathic infantile hypercalcemia early and also to prevent long-term complications.

GP57

ACUTE SUPERIOR MESENTERIC ARTERY SYNDROME – ASSOCIATION WITH GROWTH SPURTS

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Aim Superior Mesenteric Artery Syndrome is rare manifestation of duodenal obstruction arising due to external compression between the superior mesenteric artery and the aorta. The classic presentation involves postprandial abdominal pain and anorexia with weight loss. We aim to present the case of a 10-year-old female with an atypical presentation of acute Superior Mesenteric Artery Syndrome.

Method We report the comprehensive history highlighting pertinent positives and negatives, clinical examination and radiological investigations during our patient's inpatient admission.

Result Ten-year-old female, weighing 35.6 lbs and height of 153.2 cm equating to a body mass index of 15.1, with congenital adrenal hyperplasia and precocious puberty presented for elective admission for twenty-four-hour adrenal profile. On admission, patient reported worsening indigestion over past twelve months, epigastric pain and hiccups necessitating gastrointestinal consultation. Patient self-reported food to become 'stuck in the middle of her chest'. There was no associated nausea or vomiting or any recent weight loss reported. Symptoms failed to improve on a three-week course of proton pump inhibitors. A prior urea breath was negative. The patient was scheduled for barium swallow and follow through which demonstrated hold-up of the contrast in the second part of the duodenum, with the duodenal flexure in the correct position and no evidence of malrotation. After discussion as multidisciplinary conference the patient was re-admitted for elective oesophagogastroduodenoscopy. The results of which