British Paediatric Neurology Association

719 EXTRAPYRAMIDAL SIDE EFFECTS INCLUDING OCOLOGYRIC CRISIS IN TWO TEENAGERS FOLLOWING AYPTICAL ANTIPSYCHOTIC USE

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Background Extra-pyramidal side effects (EPSE) such as oculogyric crisis (OGC) are known to be precipitated by first-generation antipsychotics but are rarely seen with second generation, or atypical, antipsychotics. Atypical antipsychotics are increasingly being used for behavioural indications in children, although their side effect profile in this population remains poorly defined.

Objectives To describe EPSE, including OGC, occurring as a response to therapy with Risperidone and Aripiprazole, two atypical antipsychotics.

Methods We present the cases of two teenagers treated with Risperidone and Aripiprazole.

Results We describe and present videos of the delayed adverse extrapyramidal responses of two teenagers to these drugs.

Conclusions We raise awareness about the presentation of extrapyramidal side effects and parkinsonism following atypical antipsychotic use in the paediatric population. This is especially important as these drugs tend to be used more in children with learning difficulties who may not be able to communicate discomfort which leads to significant distress as was seen with our patients.

720 NEUROCOGNITIVE PROFILE OF MILD PHENOTYPES OF GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME (A CASE SERIES)

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Background Classical variants of Glucose transporter type 1 deficiency syndrome (GLUT-1DS) present with microcephaly, seizures, and moderate-severe developmental delay. Non-classical variants can present later in childhood with relatively milder phenotypes - epilepsy and/or paroxysmal movement disorders. There are limited descriptions of neuropsychological profiles in GLUT-1DS, and most previous work is qualitative.

Objectives To describe the neurocognitive profile of 4 children with the non-classical form of GLUT-1DS.

Methods We present a case series of neurocognitive profiles of four patients with the non-classical form of GLUT-1DS.

Results The mean age of presentation for our four cases was 6 years; clinical features were that of absence epilepsy and paroxysmal kinesigenic dyskinesia. One patient was on a ketogenic diet, one was not due to family choice, two had tried and had not been able to continue. All four patients had low average to borderline range intellectual abilities, with Verbal IQ>Performance IQ and particular difficulties were noted with visuospatial/visuomotor skills. Numeracy skills were more affected in comparison to literacy attainments and they had new learning/encoding difficulties. A high level of psychosocial stress was seen in our cohort.

Conclusions Our case series supports previous research which suggests neurocognitive deficits in childhood onset GLUT-1DS. In addition, we document particular challenges in visuospatial skills. As we have become familiar with non-classical phenotypes of GLUT-1DS only in the past decade or so, there is a need to understand the condition better in terms of long-term outcome especially as this is a treatable metabolic disorder.

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722 SOLITARY RECTAL ULCE R SYNDROME: A RARE PRESENTATION IN PEDIATRICS

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Background Solitary Rectal Ulcer Syndrome (SRUS) is an important but often delayed diagnosis with a prevalence of 1:100,000 people per year. It is rare in children. Manifestations include rectal bleeding, pain, tenesmus and, occasionally, rectal prolapse. 26% are asymptomatic. Classically, sigmoidoscopy reveals ulceration within 10cm of the anus and histology demonstrates mucosal layer thickening with crypt distortion and lamina propria fibromuscular obliteration. It is, however, a misnomer as only 20% have a solitary ulcer with the remainder have lesions ranging from hyperemic mucosa through to broad based polypoid lesions. It’s not uncommon that patients are initially misdiagnosed as constipation or inflammatory bowel disease (IBD). The mainstay of treatment includes biofeedback though some patients require surgery for rectal prolapse. Whilst the aetiology of SRUS remains unclear hypothesised causes include ineffective straining and/or uncoordinated puborectalis contraction increasing intra-rectal pressure and causing ischaemic ulceration or local vascular trauma due to intussusception.

Objectives To highlight awareness of SRUS and how it can mimic constipation and IBD.

Methods A 9 year old female patient presented with apparent constipation, resistant to laxatives. Initial abdominal pain and passage of a type 2 stool weekly progressed to rectal bleeding and passage of mucus. A later more in depth history also identified straining and tenesmus. Over several months she was treated with escalating doses of Movicol, Sodium Picosulfate, Liquid Paraffin and Klean Prep. Treatment compliance was an issue initially. Ileocolonoscopy with rectal retroflexion detected a macroscopic solitary rectal ulcer and only isolated diffuse rectal inflammation on histology. Her faecal calprotectin was normal. A diagnosis of inflammatory bowel disease was questioned but treatment with Mesalazine foam enemas was unhelpful. She also developed dysfunctional voiding during the course of treatment with significant bladder retention. Non-compliance with intermittent catheterisation due to significant anxiety led to insertion of a suprapubic catheter. Further investigation with a colonic transit study was performed due to her ongoing requirement for high dose laxatives and found markers pooled in the rectum. A diagnosis of rectal dysynergia/dysfunctional elimination was reached. She responded to suppositories which later weaned following successful use of
Investigation and Management of Vitamin B12 Deficiency: Experience in a Tertiary Paediatric Centre

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Background Vitamin B12/Cobalamin (Cbl) is a water soluble vitamin which is found in animal products (liver, salmon, beef, egg, chicken), milk products, and fortified cereals. Breast milk is sufficient to meet the demands of 0–6 month olds as long as the mother is not herself deficient. Causes of deficiency include dietary (low protein or protein restricted diet), pernicious anaemia, small bowel surgery, inflammatory bowel disease, deficient intrinsic factor, disorders in cobalamin transport and inborn errors of metabolism. Patients can present in a variety of non-specific manifestations such as pancytopenia, lethargy, chest pain, developmental delay, and weakness thus will be seen across different specialties. B12 deficiency can have significant long-term neurological effects, and the finding mandates appropriate further aetiological investigations and immediate treatment.

Objectives This audit will review current practice across all specialties at a tertiary paediatric hospital in the United Kingdom to inform further changes required to ensure optimal practice.

Methods Our study period spanned May 2018 to May 2020. We obtained data from our biochemistry department which showed 221 results with low B12 levels. We audited a random selection of the data to see if key investigations for vitamin B12 deficiency had been conducted including: Plasma homocysteine, plasma and urine methylmalonic acid (MMA), folate, full blood count (FBC), auto antibodies, intrinsic factor (IF), genetics, cobalamin, endoscopy as well as specialist investigations such as MRI and nerve conduction studies (NCS).

We also documented the main diagnosis of the patient, suspected cause of B12 deficiency, treatment plan, and if a repeat vitamin B12 level was normal.

Results Main documented cause of low Vitamin B12 include a metabolic diet (20%) mainly those on protein restriction. Other causes include gastrointestinal causes (8%) and low maternal levels in infants (5%).

49 patients (52%) had no documented cause for their vitamin B12 deficiency, of these 15 (30%) were treated with either IM or enteral vitamin B12 and of these patients 73% had repeat blood tests. Of those who were not treated, only 21% had repeat samples taken. Of all the patients, only 43% had documentation of treatment either as supplemental drop/tablets, or an IM injection.

Haematological presentations included persistent neutropenia, low vitamin B12 with variable blood sugars, and haemolytic anaemia. Homocysteine, and MMA were sent along with FBC and folate. In 1 patient, specialist investigations were sent (autoantibodies, intrinsic factor, and genetics).

Conclusions Vitamin B12 deficiency spans multi-disciplines and is an important investigation to consider for many presentations and currently there is no set local guideline; the majority of patients are not being fully investigated or treated. There is also inadequate documentation and follow-up for these patients. Metabolic patients in particular who are on dietary restrictions such as a low protein diet should have regular levels checked and treated as appropriate due to their higher risk of deficiency. Recommendations from this audit suggest a trust guideline for the initial investigation of low vitamin B12 levels, appropriate and timely management and follow-up.