Abstracts

British Paediatric Neurology Association

EXTRAPYRAMIDAL SIDE EFFECTS INCLUDING OCULOGYRIC CRISIS IN TWO TEENAGERS FOLLOWING ATYPICAL 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.

NEUROCOGNITIVE PROFILE OF MILD PHENOTYPES OF GLUTOSE 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.

British Society of Paediatric Gastroenterology, Hepatology and Nutrition

SOLITARY RECTAL ULCER SYNDROME: A RARE PRESENTATION IN PAEDIATRICS
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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 while 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 dyssynergia/dysfunctional elimination was reached. She responded to suppositories which later weaned following successful use of...