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FAST CHANNEL SYNDROME: PRESENTATION, DIAGNOSIS AND MANAGEMENT IN AN INFANT

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Aims: To report a fast channel congenital myasthenic syndrome in an infant who presented as a floppy baby with episodes of acute apnoeas.

Methodology: Case notes from a tertiary hospital were reviewed and additional literature search was performed.

Report: A 9 week old infant was referred to a tertiary unit from a local district general hospital with a history of floppiness, recurrent feeding difficulties and intermittent apnoeas. The apnoeic episodes were associated with bradycardias and desaturations needing bag and mask ventilation on most occasions.

He had had ENT, speech and language assessment, MRI brain and a basic metabolic workup at the time of transfer.

In our hospital he was further assessed by various professionals and investigated accordingly for a floppy baby syndrome.

At 4 months of age he was admitted to paediatric intensive care unit following a respiratory arrest. On this occasion was as noted to have ptosis and opthalmoplegia which was more obvious He had a Tensilon test performed which was positive (video clip available). Following this he had nerve conduction tests and electro myography which confirmed a myasthenic syndrome.

DNA testing revealed the diagnosis of a fast channel congenital myasthenic syndrome.

He has responded well to pyridostigmine, but continues to have infrequent intermittent respiratory crises. Management issues are discussed.

Conclusion: This is the first case of fast channel congenital myasthenic syndrome in Northern Ireland. Ptosis and opthalmoplegia may not be obvious on initial examination, all floppy infants should be explored for congenital myasthenic syndrome.

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THE FIRST TURKISH CASE OF GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME (GLUT 1D) WITH MOLECULAR STUDIES

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Glucose transporter type1 (GLUT-1) deficiency syndrome (OMIM 606777) is a autosomal dominantly inherited complex neurologic disorder that causes severe learning difficulties, ataxia, seizures, acquired microcephaly and developmental delay. We present a first Turkish case with GLUT1-Deficiency syndrome which is confirmed with molecular studies and additional four cases described. Children presenting with a clinical phenotype consisting of a refractory seizure disorder, ataxia and developmental delay should prompt the consideration of Glucose transporter 1 deficiency syndrome.

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DEMOGRAPHICS AND DIAGNOSES - A PAEDIATRIC NEURODEVELOPMENTAL CLINIC'S EXPERIENCE

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Background and aims: Children from ethnic minorities are likely to receive a diagnosis of autism 1.4 years later than non minority children and are at increased risk of developmental delay. This study aims to identify referral patterns, diagnoses, family developmental history, and nationality presenting to a paediatric neurodevelopmental clinic.

Methods: A review of medical records of children referred to a paediatric neurodevelopmental clinic from 2007 to 2009 was performed.

Results: 342 children presented. 218 (64.17%) were Irish and 101 (29.5%) were from a minority ethnic group. Of this group 54 (53.5%) were African. The ethnicity of 23 (6.7%) was unknown. 118 children (34.5%) had a primary diagnosis of

speech and language delay and 106 (31%) had a diagnosis on the autistic spectrum(ASD). 12 (3.5%) had behavioural problems and 16 (4.68%) children had a primary diagnosis of intellectual disability. 79 (23.1%) children had cognitive impairment. 47 (13.7%) are awaiting completion of assessment. 22 (40.1%) of the 54 African children, and 42 (41.25%) of the total ethnic minority cohort had an ASD diagnosis. 72(33%) of the Irish children had an ASD diagnosis.

Conclusions: This clinic has a disproportionate number of referrals from ethnic minority backgrounds. The high number of referrals of African children is interesting, especially considering that the African community comprises 0.85% of the Irish population, and that 5.6% of births in 2004 were to African mothers. Children presenting from ethnic minority backgrounds were more likely to have an ASD diagnosis than the Irish children, a feature which requires investigation.

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TRANSANAL IRRIGATION IS A VALID AND SAFE APPROACH FOR NEUROGENIC CONSTIPATION IN MYELOMENINGOCELE CHILDREN

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The aim of this study is to investigate if transanal irrigation is a valid approach for neurogenic constipation in children with myelomeningocele.

100 patients (60 boys and 40 girls; aged 8-20 y) with neurogenic bowel dysfunction were treated with transanal irrigation for six months. A questionnaire on the effects on bowel disturbances, self-management and quality of life related to neurogenic constipation was completed before and after the trial.

At the end of the trial 66 % (66/100) of patients referred a successful in constipation and 70% (70/100) regarding faecal incontinence. Questionnaire scores before and at termination of the study showed that neurogenic bowel dysfunction total score (range 0-47, 47= severe bowel dysfunction) was 17.5 (5.2) versus 8.5 (4.3) (P = .001), frequency of fecal incontinence (range is 0-13, 13= daily) was 5.5 (1.2) versus 1.3 (1.7) (P = .01) and degree of general satisfaction (range 0-10, 10= high satisfaction) was 3.0 (2.4) versus 7.7 (1.5) (P = .001).

We observed a moderate reduction of number of urinary tract infections during treatment particularly regarding E.Coli infections: 24 total urinary tract infections (18 by E. Coli) in 6 months before versus 12 (9) during treatment (P< 0.01). No changing regarding urodynamic parameters were found. No severe adverse effects were reported and treatment was been well tolerated by younger children too.

Transanal irrigation is a valid approach for managing of neurogenic constipation in fact it improves bowel disturbances, quality of life and seems to reduce urinary tract infections.

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IMPROVING CLINICAL CARE BY ROLLING AUDIT IN A DISTRICT GENERAL HOSPITAL EPILEPSY CLINIC

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Aims: Dedicated paediatric epilepsy clinics are increasingly recognised as good practice within DGH's. We aimed to gather demographics and audit current practice against 2004 NICE guidelines in the context of a data set encouraging clinicians to use the ILAE approach to paediatric epilepsy, forming the basis for a clinically relevant database.

Method: Proforma were completed for all children attending the epilepsy clinic. Demographics and clinical outcomes were measured and audited against NICE guidelines.

Results: 110 children attended clinic of which 83 had epilepsy. 55% were male. 18% presented < 2 years old. Common seizure types were generalised tonic clonic, absence and focal motor. Overall epilepsies were classified as generalised (37%), focal (48%) and encephalopathic (14%). A specific epilepsy syndrome was identified in 86%. Common epilepsy syndromes included symptomatic focal epilepsy, generalised tonic clonic and BECTS. Symptomatic or probably symptomatic aetiology was identified in generalised (29%), focal (68%) and encephalopathic epilepsy (100%). Associated co-morbidities were common (60%). Significant learning difficulties occurred in generalised (19%), focal (25%) and encephalopathic epilepsy (91%). Paediatric neurology was involved in 46%. AED choice was appropriate in 99%. Monotherapy was achieved in 65% requiring AEDs and 14% were on > 2 AEDs. EEG was performed in 100%, MRI in