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Aicardi-Goutières syndrome: a genetic syndrome mimicking congenital infection – a description of two new cases

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Aicardi-Goutières syndrome (AGS) is a progressive encephalopathy, with a recessive autosomal pattern of inheritance that has its onset in the first year of life and is characterized by acquired microcephaly, basal ganglia calcifications, white matter abnormalities, chronic lymphocytosis and raised interferon-alpha in cerebrospinal fluid. Many of these features overlap with those of an intrauterine infection and can therefore lead to the wrong diagnosis.

Here we describe two siblings, a brother and a sister, with clinical features initially suggestive of a congenital infection but with negative serological TORCH analysis. Further testing confirmed AGS.

AGS is an autosomal recessive syndrome that can mimic congenital infection. It is important to recognize because of the progressive nature of the syndrome and the risk of recurrence in families with affected children.

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Special educational needs teams (SENT) project in schools – a model of service delivery

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Aims: Special Educational Needs Teams (SENT) project was initiated in October 1998 through joint collaboration of child support services within the Education and the Health Divisions. The aim was to implement the project as a pragmatic response to the need for a co-ordinated approach in the support of children in schools in order to overcome some of the problems of poor interagency liaison and lack of community resources.

Methods: Work was initially piloted in two State primary schools and in subsequent years, the number of participating schools varied from 4-10 schools/year. Team structure, method of work and evaluation procedures were developed in order to provide a basis for individual case studies, assessing whole school needs and developing a plan

Results: Over the past 6 years, SENT provided support to 38 Mainstream schools. An average of 7 meetings /school / year was held and an average of 14 cases was reviewed / team. The team liaised with a total of 11 different agencies. Predominant reasons for referral included emotional / behavioural difficulties, speech and language problems, learning difficulties and psychosocial problems. Whole-school needs evaluation varied but a recurring theme included the need for a resource room, social support and parental and teacher information.

Conclusion: Feedback received from schools regarding the project has been generally positive. There was a consensus feeling that SENT has enhanced inter-agency collaboration and offered support not only to children and their parents but also to the whole school environment.

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Griscelli syndrome - the commonest cause of Haemophagocytic Lymphohistiocytosis in Maltese children

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Aims: The presentation will describe the clinical features, laboratory findings, diagnostic criteria and treatment modalities of children with the Griscelli syndrome and Haemophagocytic Lymphohistiocytosis who presented to the Department of Paediatrics in Malta from 2005 to 2006.

Methods: Three short case presentations will be followed by a description of the Griscelli syndrome and Haemophagocytic Lymphohistiocytosis.

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DC cardioversion in a case neonatal atrial flutter

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Flutter is very rare in the paediatric age group, particularly in the neonatal period. We present a newborn with atrial flutter in the absence of structural heart disease. Tachycardiomyopathy was already present i.e. cardiac dilatation and failure due to excessive heart rate – 400/min atrial rate with 2:1 block producing a ventricular rate of 230/min. Medical treatment failed and DC shock reverted the rhythm to sinus with rapid normalisation of cardiac function.

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Transmission of ring chromosome 21 from a phenotypically normal mother to her trisomic daughter

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Introduction: Ring chromosome 21 is a rare abnormality giving rise to a wide variety of phenotypes. The origin of ring 21 chromosome also varies and may include deleted or duplicated material.. Most cases arise de novo, but some are transmitted through generations. We analysed the mechanism of origin of mosaic trisomy 21 that included an additional ring 21 chromosome of maternal origin.

Methods: G-banded Karyotype and Telomeric FISH analysis.

Results: The baby showed some phenotypic features of Down syndrome. Her karyotype showed a mosaic 46,XX / 47,XX+r21 The ring chromosome was present in the trisomic cell line only, while the 46 XX cell lines all contained a pair of normal chromosome 21. Her mother, a 35 year old lady with a normal phenotype and a record of consistently poor school performance had a non-mosaic karyotype 46,XX -21, + r21. FISH analysis showed the presence of a single centromere, and loss of 21q telomeres in the maternal ring chromosome 21 and in all trisomic cells of the child.

Conclusions: The conceptus originated as a maternal non-disjunction giving rise to a trisomy 21 zygote. Mosaicism arose post-zygotically by trisomic rescue in some blastomeres. Ring chromosome formation involved fusion of the distal ends of the long and short arms of chromosome 21 with consequent loss of 21q telomeres. The telomeric loss apparently had very small effects on the phenotype resulting only in intellectual impairment in the mother. Evidence indicates that ring chromosome 21 also predisposes to non-disjunction.

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Therapeutic drug monitoring of lamotrigine in a paediatric population

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Aims: Therapeutic drug monitoring is important for drugs that exhibit inter-individual variability in pharmacokinetics, and where drug-drug interactions, concurrent disease or age alters the kinetics of that drug. This is of particular importance in a chronic neurological condition such as epilepsy. In this study, the value of therapeutic drug monitoring for lamotrigine, a novel antiepileptic drug, was investigated in a group of paediatric patients.

Methods: Plasma lamotrigine levels at steady state (mean ± S.D.) in epileptic patients were thus measured using this novel analysis.