MEASURING THE NUTRITIONAL STATUS OF CHILDREN WITH JUVENILE IDIOPATHIC ARTHRITIS USING THE BIOELECTRICAL IMPEDANCE METHOD

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Aims: To assess the nutritional status of children with Juvenile Idiopathic Arthritis (JIA) using anthropometric measurements and bioelectrical impedance.

Methods: Twenty-two consecutive JIA patients, 7 pauciarticular, 15 polyarticular, attending the rheumatology clinic at Booth Hall Children's Hospital were compared to twenty-two age and sex matched controls attending the accident and emergency department at the same hospital. There were no patients with systemic onset JIA in the cohort. Height, weight, head circumference and skinfold thickness at four sites (biceps, triceps, subscapular and suprailiac) were measured. Regression equations were used to calculate the percentage body fat of weight and arm muscle circumference. In addition bioelectrical impedance measurements were made using a Holtain body composition analyser. These measurements were then used to calculate the total body water, which could be used as an indirect estimate of the lean body mass.

Results: 22.7% of the JIA patients were below the 3rd centile for height, 18.1% had a weight less than the 3rd centile. Mid arm circumference was below the 5th centile in 36.4% of the patients. Knowledge with polyarticular disease showed significantly more signs of malnutrition than pauciarticular. In the polyarticular group comparison with controls revealed significant p values for reduction in height (0.047), weight (0.045), mid-arm circumference (0.002), arm muscle circumference (0.012), percentage body fat (0.008) and total body water (0.031).

Conclusions: Bioelectrical impedance is a useful adjunct to anthropometric measures in assessing nutritional status in JIA.

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GROWTH HORMONE THERAPY IN CHILDHOOD-ONSET SYSTEMIC LUPUS ERYTHEMATOSUS

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Introduction: Growth retardation is a frequent and serious complication of childhood arthritis, particularly in those with systemic disease. Growth hormone (GH) treatment has been used in this condition but experience is limited.

Aims and methods: To audit the use of GH therapy in childhood arthritis over a 10 year period and to assess the effects of steroid treatment on growth response.

Results: 10 children received growth hormone therapy in 11 treatment episodes. 8 had systemic onset JIA, 1 had Sjögren's syndrome and one had polyarticular with Glycogen Storage Disease. Treatment was commenced at 6.5–13 years age and continued for a mean of 3.5 years (2–7 years). Mean GH dose was 0.86u/kg/week (range 0.73–1.14). All had significant growth retardation with a mean height SDS of −3.96 (range −2.41 to −5.59) and all were pre-pubertal prior to GH treatment. The mean pre-treatment height velocity (HV) was 2.42cm/yr with the mean first year treatment HV being 4.63cm/yr (range 1.4 to 8.0) and second year 4.3cm/yr (range 0.5 to 10.9). 6 children showed an improvement in height SDS with 2 entering the normal range. There was an inverse relationship between change in HV in the first year and prednisolone dose, with those receiving the equivalent of 5mg/day or less showing a better response.

Conclusions: Although most children showed an improvement in height velocity the response was highly variable and the change in height SDS was only small. The data suggests that children on a prednisolone dose >5mg/day have a limited response to GH therapy.
Clinical Genetics

G176 CURING TWO BIRDS WITH ONE STONE: A CASE OF FAMILIAL HIBERNIAN FEVER

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A six-year-old indigenous Scottish girl was seen on two occasions in a local district general with febrile episodes. The first episode was attributed to an E.Coli found on blood culture, but the second episode remained unexplained and resolved spontaneously.

When a further episode of unexplained fever occurred, referral was made to the rheumatology service at the Royal Hospital for Sick Children for further assessment. Investigation revealed no infective, immunological, rheumatic, or malignant aetiology. However, the ESR and CRP were markedly elevated.

On further questioning, the patient’s father described identical episodes of fever and chest pain occurring every few months for 30 years. He had been investigated repeatedly but with no cause found. This history suggested a periodic fever syndrome. Blood was sent from both for DNA analysis to identify recognised mutations seen in Familial Mediterranean Fever and Familial Hibernian Fever (FHF).

The presence of a mutation in the Tumour Necrosis Factor Gene was identified in both father and daughter and so a diagnosis of FHF was made.

This is an autosomal dominant condition previously described in three Irish families. It runs a benign course with individual episodes treated with oral steroids.

G177 EXTREME VARIABILITY OF EXPRESSION IN A FAMILY WITH THE SONIC HEDGEHOG MUTATION

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Introduction: Holoprosencephaly is a clinically variable and genetically heterogeneous CNS malformation. Alobar Holoprosencephaly is the most severe form and usually associated with a poor prognosis. At the milder end of the holoprosencephaly spectrum microcephaly, hypotelorism, and single centre maxillary incisor may be recognised. Currently, four genes have been identified for this condition. These include the Sonic Hedgehog (SHH) gene on chromosome 7q36, which is thought to be responsible for a large proportion of autosomal dominant holoprosencephaly.

Results: We report an index case with classic alobar holoprosencephaly and a SHH mutation and six members of his family with this mutation across two generations with a broad range of clinical presentation.

Four daughters in the same family carry the SHH mutation and two of these children who also carry the mutation. Members of this family who are identified as having the mutation have a wide range of clinical presentation. All have microcephaly, two have midline defects including cleft lip and palate and hypospadias, one has iris coloboma, and four have significant learning difficulties. Two individuals presented with hyperactivity, poor concentration and microcephaly. MRI findings are presented in three of the children and the different phenotypes presenting with the same mutation are discussed.

Conclusion: The presentation of microcephaly, hypotelorism and subtle midline facial anomalies in a child should alert the physician to the diagnosis of holoprosencephaly.

G178 ARC SYNDROME: AN EXPANDING RANGE OF PHENOTYPES

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Aim: To describe the clinical phenotype in infants with ARC syndrome, the association of arthrogryposis, renal tubular acidosis and cholestasis.