Method: A case report of a 13 year old girl who died of Juvenile onset Huntington’s Disease in a children’s hospice.

Results: This child died 3 years after her diagnosis and 8 years after she first presented with symptoms of this rare dominantly inherited condition, proving a challenge for both diagnosis and management. Her major symptoms in the terminal phase included pain, dystonia and muscle spasm, agitation and excessive respiratory secretions. 30% of the drugs that were required to control her symptoms were used ‘off licence’. There is a lack of evidence-based treatments in Palliative Care, and a need to use drugs ‘off licence’. The report also considers other ethical issues that arise in Paediatric Palliative Care, such as gastrostomies for children with neurodegenerative conditions.

Conclusion: Paediatric Palliative Care is usually discussed with reference to children with malignant conditions, however far more children die from non-malignant conditions and this report offers the opportunity to consider the palliative care management of a child with a neurodegenerative condition.

G191 CUTANEOUS GRANULOMATA AS A PRESENTATION OF IMMUNE DYSREGULATION


Cutaneous granulomata can occur as a component of a number of diseases including typical and atypical mycobacterial infections, sarcoidosis, granuloma annulare or as a manifestation of immunodeficiency.

We describe four cases of granulomatous skin disease in children. All four presented with skin lesions before the age of eighteen months. The cutaneous lesions in each child were stereotyped, manifesting as red/purple papules with central atrophy and scarring, affecting the face and limbs but sparing the trunk. Biopsy showed intradermal granulomata with histological features not consistent with mycobacterial infection. Furthermore, lesions did not respond to atypical or typical antimycobacterial agents.

At presentation all children had overtly normal immune function. All four subsequently developed abnormalities in quantitative and/or qualitative aspects of immunity: hypogammaglobulinaemia, low numbers of CD8 positive T cells and overt immune dysregulation with life threatening haemophagocytic syndrome.

All cases responded to oral corticosteroid therapy but the patients developed significant side effects and required steroid sparing agents. Responses to the latter, which included intravenous immunoglobulin and azathioprine, were variable and all alternative agents were less efficacious than steroids.

Granulomatous skin lesions are an important early sign of possible immunodeficiency, and long term dermatological and immunological monitoring is recommended.

G192 THE PARENTAL VIEW OF ECZEMA AND ITS MANAGEMENT

J. Levy, H.M. Goodyear. Birmingham Heartlands Hospital, UK

Aims: To assess a range of parental concerns and perceptions regarding eczema. The areas concentrated on were 1) adherence to management regimes 2) effect of eczema on the sleep of children and carers 3) satisfaction with the medical care 4) parental attitudes towards steroid creams 5) attitudes towards, perceived curative qualities and use of “alternative therapies” 6) parental perception of their child’s allergies and food intolerances.

Methods: Questionnaires were distributed to parents of eczema- tous children at outpatient appointments and were completed whilst in the waiting area.

Results: 128 questionnaires completed. 1) Strict adherence to treatment rose from 38% of respondents to 83% when symptoms worsened. 2) 70% of children lost sleep each week (median hours lost = 8.5). 60% of carers sleep was affected (median hours lost = 10). 3) 79% of parents were satisfied with their child’s care. 14% of parents felt that their expectations for the consultation were not met. 4) 22% of parents did not perceive any effect of steroid creams. 48% were less happy to give their children steroid creams than other medicines. 58% gave it “exactly as prescribed”. 5) 78% of parents believe that alternative therapies could help eczema. 6) 66% believed that food affects eczema. 77% believed that allergies affect eczema. 47% believed that allergies cause eczema.

Conclusions: Parents harbour misconceptions about eczema, they do not treat it consistently or adequately and have faith in alternative therapies. Unless these issues are addressed, there will continue to be a high rate of non-compliance with eczema treatment.

G193 A CASE OF TRICHORHINOPHALANGEAL SYNDROME IN MONOZYGOYTIC TWINS

T. Dawson, A. Heggarty, S.J. Rose, H. Goodyear. Birmingham Heartlands Hospital, UK

Patients with Trichorhinophalangeal Syndrome commonly present to dermatology clinics with curly, sparse, brittle, slow growing scalp hair.

www.archdischild.com
There are some components of this condition, which if identified can be treated. It has been recognised that these patients are often subjected to aesthetic or plastic procedures prior to diagnosis. We describe female, twelve-year-old monozygotic twins diagnosed with Trichorhinophalangeal Syndrome Type 1 at a dermatology clinic and referred to our endocrinology clinic for short stature.

The estimated final height for each of these girls was below the 5th centile. GH treatment has been shown to be useful in other bony dysplasias. We postulated that growth hormone (GH) may accelerate growth velocity resulting in an increased final height. As both girls were in puberty, pubertal progress was halted with GNRH analogue and each was prescribed the dose of GH consistent with normal GH levels (as used in Turner Syndrome).

Over a two year period these girls have maintained a growth velocity between the 80th and 90th centiles, over and above the initial growth spurt expected in those beginning GH treatment. It appears that increased growth velocity may be gained by commencing GH therapy in patients with Trichorhinophalangeal Syndrome Type 1. This can improve their final height outcome. In those children who have started puberty, benefit may be gained by stopping puberty first. This stresses the importance of identifying patients with bony dysplasias in the dermatology clinic where they are most commonly seen and diagnosed.

Allergy, Immunity, and Infection

**G194**  THE PRO- AND ANTI-INFLAMMATORY CYTOKINE PROFILE IN CHILDREN WITH MENINGOCOCCAL DISEASE

E.D. Carrol, A.P.J. Thomson, K. Mobbs 1, J.A. Sills, C.A. Hart 1. Institute of Child Health, RLCH NHS Trust, Alder Hey, Eaton Road, Liverpool L12 2AP; 2Dept of Medical Microbiology, University of Liverpool, Daulby Street, Liverpool, L69 3GA, UK

**Introduction:** In meningococcal disease (MCD) there is early activation of both pro- and anti-inflammatory cytokines triggered by the release of endotoxin. Some studies of MCD have claimed that an anti-inflammatory cytokine profile is associated with a fatal outcome, so contra-indicating pro-inflammatory cytokine inhibition therapies. Other studies have demonstrated down-regulation of pro-inflammatory cytokines and up-regulation of anti-inflammatory cytokines and suggested this as a protective strategy in MCD.

**Aims:** To determine whether an anti-inflammatory cytokine profile in MCD was associated with an increased risk of severe disease or death.

**Methods:** A total of 112 children with MCD were prospectively studied. Plasma concentrations of interleukin-1 receptor antagonist (IL-1Ra), interleukin-6 (IL-6), and tumour necrosis factor-α (TNF-α) were assayed on admission. Severe disease was defined as a Glasgow Meningococcal Septicaemia Prognostic Score (GMSPS) of ≥8.

**Results:** A high IL-1Ra:TNF-α ratio (>20) was associated with less severe disease (p=0.014). There was a trend in favour of an association between lower IL-1Ra:TNF-α ratios and death, but this was not significant (p=0.283). A lower proportion of children with a high IL-1Ra:TNF-α ratio developed septic shock (6%) than those with a low ratio (38%), p<0.0005. In children with a high IL-1Ra:TNF-α ratio the relative risk of severe disease was 0.63 (95% CI 0.42–0.91), odds ratio 0.39 (95% CI 0.17–0.89). A high IL-1Ra:IL-6 ratio was not significantly associated with severity of disease or risk of death.

**Conclusions:** An anti-inflammatory profile with a high IL-1Ra:TNF-α ratio appears to be associated with a favourable prognosis and supports the concept that early IL-1Ra therapy with TNF-α inhibition might be beneficial.

**G195**  TaqI POLYMORPHISM IN THE 3’ FLANKING REGION OF THE ALPHA-1 ANTITRYPSIN GENE IN CHILDREN WITH MENINGOCOCCAL SEPTICAEMIA

D. Hodge 1, P.C. Holland 1, A. Finn 1, D. Jury 1, D. Bonthron 1. 1Department of Paediatrics and Child Health, Leeds General Infirmary, Leeds, LS1 3EX; 2Sheffield Institute for Vaccine Studies, Children’s Hospital, Sheffield S10 2TH, UK

**Introduction:** As part of the inflammatory response in patients with meningococcal septicaemia, large numbers of neutrophils are stimulated, releasing elastase, a proteolytic enzyme which causes vascular endothelial injury which is normally counteracted by the inhibitor α-1-antitrypsin (A1AT). It is hypothesized that a polymorphism in a Taq I restriction enzyme recognition site, 3’ to the gene for A1AT, results in an inability to produce sufficient A1AT during the acute phase response, and therefore increases susceptibility to septic shock.

**Method:** Blood samples were collected from paediatric patients with meningococcal septicaemia (n=112) and a control group of paediatric patients (n=154). DNA was extracted and the TaqI recognition site was amplified using PCR and the fragments digested using TaqI restriction enzyme.

**Results:** Almost twice as many meningococcal patients had the TaqI polymorphism than in the control group (20.5% and 11.6% respectively, χ² =3.89, p<0.05).

**Conclusions:** The results provide further evidence of a link between α-1-antitrypsin and susceptibility to meningococcal septicaemia. Further investigations are needed to elucidate the exact effect of the TaqI polymorphism.