Rheumatology and dermatology joint session

G197 CONGENITAL INSENSITIVITY TO PAIN PRESENTING AS DIGITAL NECROSIS

K Neuling, H Goodyear. *Heart of England NHS Foundation Trust, Birmingham, UK*

Introduction: Congenital insensitivity to pain is due to a number of inherited disorders that are associated with sensory and autonomic dysfunction. Herpetic whitlow is a herpes simplex virus type 1 or 2 infection of the fingers characterised by erythema and painful vesicles. It is a self-limiting condition in the immunocompetent individual and is not usually associated with necrosis or scarring. A 13-month-old boy who developed a gangrenous fingertip following presumed herpetic skin infection is presented.
Case report: He presented with a 1-month history of changes affecting the thumb and tip of the index finger that were black and necrotic. The history was one of vesicular lesions with surrounding erythema. There was also marked ulceration and oedema of the lower lip and tip of the tongue. He showed no signs of discomfort despite the dramatic skin changes. His parents had previously noted that he did not cry in response to falls or when he was immunised. He is the first child of consanguineous parents with no significant family history. His development is normal and he is felt to have normal sweating. Over a period of 2 weeks, the necrotic index finger tip auto-amputated. Other skin changes improved following intravenous aciclovir and antibiotics. Blood urate levels were normal. Neurophysiological studies showed no evidence of significant peripheral neuropathy. Congenital insensitivity to pain, hereditary sensory and autonomic neuropathy (HSAN), is considered the most likely diagnosis in this patient. His symptoms are not completely characteristic, however, of previously described types.
Conclusions: The diagnosis of HSAN is based on clinical features and the genetic defect varies according to type. It is important to think about underlying disorders in children, particularly when a child with normal immunity presents with unusually severe and destructive skin lesions.

G198 A CASE OF "SKIN AND BONES": THE SYNOVITIS, ACNE, PUSTULOSIS, HYPEROSTOSIS AND OSTEOMYELITIS SYNDROME

R Frost, L Jindal, T Winter, R Logan, J Hughes, N D'Souza. *Cardiff University, Cardiff, UK*

Introduction: SAPHO syndrome (synovitis, acne, pustulosis, hyperostosis and osteomyelitis) is a rare constellation of skin, joint and bone disease first described by Chamot in 1987. There are characteristic clinical and radiological features and the underlying pathological process is a sterile osteitis. Our patient demonstrates the typical features of the syndrome.
Case report: A 15-year-old boy presented with fatigue, anorexia, ankle pain and chest wall discomfort and swelling. On examination he was febrile; he had nodulocystic acne over his face, chest and back. There was a tender swelling over the left sternoclavicular joint with limited shoulder abduction and mild swelling over the lateral aspects of his ankles. Investigations showed: CRP 104 mg/l, ESR 102 mm/h, WBC 16.8×10^9, neutrophil count 13.2×10^9, rheumatoid factor negative and blood cultures negative. *x*-Ray of the left clavicle showed a marked periosteal reaction, confirmed with MRI scan. *x* Ray ankles were normal. Bone scan demonstrated increased uptake at the medial end of the left clavicle and the metaphyseal region of both ankles. Treatment with high-dose oral steroids followed by an incremental dose of isotretinoin resulted in a rapid reduction in inflammatory markers. The chest swelling and joint pain subsided gradually and his acne improved over 6 months.
Conclusions: Recognising the association of acute joint symptoms with acne, a common condition in the adolescent population is important to avoid unnecessary investigations and to help in the treatment and long-term management of this rare group of patients.

G199 HAIR-THREAD TOURNIQUET SYNDROME MIMICKING NON-ACCIDENTAL INJURY: A REPORT OF TWO CASES AND REVIEW OF THE LITERATURE

[1]V Palanivel, [2]MA Anjay, [2]J Haider, [2]S Nirmal. *[1]Addenbrooke's Hospital, Cambridge, UK; [2]James Paget University Hospital, Great Yarmouth, UK*

Objective: Hair-thread tourniquet syndrome is an unusual condition and is often confused with non-accidental injury (NAI). We report two cases of hair-thread tourniquet syndrome presenting as NAI followed by a brief review of literature.
Methods: Case reports and literature review.
Results: Two children were referred with suspicion of non-accidental injury with circumferential wounds affecting the digits. The first child, an 11-week-old boy, had a cut encircling the right third toe at the level of the distal interphalangeal joint. Swelling and erythema were present distal to the lesion. On further assessment the inside of the child's socks was noticed to have loose threads. Her grandmother reported pulling out a thread from the lesion sometime earlier. The other child, a 2-month-old girl, presented with a similar lesion on the right thumb. She was noticed to be playing with mum's hair frequently. Examination under an operating microscope revealed a hair within the lesion, which was removed. Both children recovered uneventfully. Literature review revealed a total of 218 reported cases with 266 appendages involved. Penis is the most common appendage involved, followed by toe. Hair is the most common offending fibre. The age range is from 2 days to 84 years.
Conclusions: Hair-thread tourniquet syndrome is a rare condition affecting the appendages, mostly in infants and young children. This can involve fingers, toes and sometimes genitals, when they are accidentally strangulated by a hair or thread. This leads to obstruction of the circulation resulting in swelling, ischaemia and eventually necrosis. Early diagnosis and treatment is essential to prevent permanent damage. Surveys of health professionals by Biehler *et al*[1] revealed that a child welfare worker would interpret this condition as child abuse and make a referral to the appropriate authorities more frequently than physicians or public health nurses. The observed difference is the result of a lack of familiarity with this condition. In both our cases, NAI was suspected, but was excluded after detailed assessment. Awareness of this interesting condition, coupled with a thorough history, will prevent misdiagnosis as NAI.

1. **Biehler JL,** Sieck C, Bonner B, *et al*. A survey of health care and child protective services provider knowledge regarding the toe tourniquet syndrome. *Child Abuse Neglect* 1994;**18**:987–93.

G200 DEGOS DISEASE: A RARE OCCLUSIVE VASCULOPATHY MIMICKING POLYARTERITIS NODOSA

[1]A Kelly, [2]P Riley, [3]N Laycock, [1]N Sebire, [2]M Newbould, [1]N Klein, [1]J Harper, [1]P Brogan. *[1]Great Ormond Street Children's Hospital NHS Trust, London, UK; [2]Royal Manchester Children's Hospital, Manchester, UK; [3]Blackpool, Fylde and Wyre Hospitals NHS Foundation Trust, Blackpool, UK*

Aims: We present a case of Degos disease, a rare occlusive vasculopathy with fatal outcome. We highlight this case as a rare but important mimic of vasculitis with no effective treatment and which is often fatal.
Case: A 14-month-old girl was referred for opinion and diagnosis. A first child of a non-consanguineous relationship she was born at

term with a birth weight of 3.3 kg. Two skin lesions were noted at birth. These increased in number from 2 months of age onwards. They were approximately 0.5 cm diameter papules present on the trunk, limbs and scalp with a necrotic pale ''porcelain'' scarred centre. At one stage these had been mistaken for cigarette burns. Skin biopsy showed a perivascular, predominantly mononuclear inflammatory infiltrate around the small deep dermal vessels, with a lichenoid reaction affecting the adnexae. She had chronic diarrhoea, abdominal distension and severe failure to thrive (weight <0.4th centile). Bilateral ptosis was noted at 9 months of age. CT of the brain at 11 months showed areas of calcification in the left frontal area, around the third ventricle and punctate calcification in the right parietotemporal area. Inflammatory markers were normal, and extensive screening for congenital infection and autoimmune disease was negative. Aged 12 months she became acutely unwell with severe abdominal distension and peritonitis. Laparotomy revealed copious ascitic fluid and multiple pale areas on her bowel wall, but no bowel perforation. Peritoneal swabs were negative on culture. The striking appearance of typical skin lesions, ptosis, ascites with patchy bowel ischaemia, and CNS involvement suggested the clinical diagnosis of Degos disease. She was commenced on antiplatelet doses of aspirin, low molecular weight heparin and nifedipine. Despite this, and a subsequent therapeutic trial of corticosteroids, she continued to deteriorate and died at the age of 16 months. Post mortem confirmed the diagnosis of Degos disease with multisystemic occlusive vasculopathy.

Conclusions: Degos disease is a rare occlusive vasculopathy with no effective treatment and frequently fatal. The non-specific perivascular inflammatory infiltrate on skin biopsy raised the possibility of vasculitis such as polyarteritis nodosa. Importantly, however, corticosteroids are thought to worsen the occlusive vasculopathy associated with Degos disease, and immunosuppression is ineffective. Thus we now add Degos disease to the list of vasculitis mimics presenting in young children.

G201 DEGOS DISEASE: A RARE VASCULAR OCCLUSIVE DISEASE CAUSING SUBDURAL COLLECTIONS AND SKIN LESIONS

[1]S Omar, [2]C Moss, [1]S Hackett, [2]E Wassmer, [2]G Debelle. *[1]Birmingham Heartlands Hospital, Birmingham, UK; [2]Birmingham Children's Hospital, Birmingham, UK*

The diagnosis and management of non-accidental injury (NAI) is difficult and contentious with several recent high-profile cases. We report a case where differentiating between NAI and organic disease was particularly challenging.

Case report: A 6-month-old boy presented with a 1-week history of being non-specifically unwell. He also had mild eczema and scattered skin ''ulcers''. He was discharged with follow-up but re-presented 3 days later with right-sided focal seizures. Examination showed an afebrile, irritable infant with disconjugate eye movements but no fundal haemorrhages. There were several 7–10 mm skin lesions with a red margin and white, atrophic centre on the trunk, limbs and scrotum. A CT brain scan revealed bilateral subdural fluid collections and a subdural peritoneal shunt was inserted. MRI confirmed no intracerebral lesion. Cerebrospinal fluid protein level was raised at 9.85 g/l (normal range <0.25 g/l). Bilateral subdural collections in a non-mobile infant and skin lesions resembling contact burns raised the possibility of NAI. Differential diagnosis included a vasculitis; however, inflammatory markers were normal and skin histology showing ulceration with a vasculopathic reaction and leucocytoclasis but no fibrinoid necrosis did not confirm this. The child's condition initially stabilised, but then deteriorated rapidly with recurring seizures and cranial nerve involvement. Serial photographs showed no resolution of the skin lesions and further investigations were undertaken. Skin and dura biopsies showed arterioles with non-inflammatory transmural infarction and thickened proliferative vascular endothelium confirming Degos disease. He died 8 weeks after admission with multiple cerebral infarctions.

Discussion: Degos' malignant atrophic papulosis, is a rare non-inflammatory vascular occlusive disease of unknown aetiology usually seen in adults. In a third of patients the disorder is confined to the skin but the bowel and neurological system can be involved. There are no proven treatments and systemic involvement is usually fatal. This case reminds us that in cases of suspected child abuse when there are unusual features, any diagnosis suggested, however unlikely, must be considered carefully, and the literature reviewed thoroughly, before diagnosing NAI.

G202 WEGENER'S GRANULOMATOSIS PRESENTING AS HENOCH–SCHOENLEIN PURPURA!

C Johnson, R Murphy. *Queens Medical Centre, Nottingham, UK*

Wegener's granulomatosis is a multisystem disease diagnosed by a triad of necrotising granulomatous vasculitis affecting both arteries and veins and focal granulomatous change in the respiratory and renal tracts. The condition is very rare in children but if untreated can be fatal. Here we describe the case of a 13-year-old girl who presented with a vasculitic rash that was initially misdiagnosed as Henoch–Schoenlein purpura (HSP). She initially presented via A&E with a vasculitic rash on her legs and a sore mouth. However, the lower limb vasculitis was atypical for HSP with a solitary deep ulcer that was slow to heal. She also complained of nasal crusting for several months and the GP referred her to ENT who considered a diagnosis of Wegener's granulomatosis. She was therefore referred to the joint dermatology/rheumatology clinic. By this time her condition had deteriorated. The leg ulcer and nasal crusting persisted but in addition she was significantly anaemic, short of breath, lethargic and had proteinuria. Appropriate investigations were carried out including a chest *x*-ray showing bilateral perihilar opacification and a renal biopsy showing granulomatous vasculitis. ANCA PR3 was positive. The skin biopsy did not show any granulomatous change. A diagnosis of Wegener's granulomatosis was thus confirmed. The patient was immediately commenced on pulsed cyclophosphamide and methylprednisolone. She continues to do well with methotrexate 20 mg weekly and cyclophosphamide every 3 months. Wegener's granulomatosis is rare in children but should be considered if there is an area of deep vasculitis on the skin that is slow to heal as this would be an atypical presentation of HSP.

G203 AN UNUSUAL PRESENTATION OF SYSTEMIC LUPUS ERYTHEMATOSUS

D Hawley, R Murphy. *Queen's Medical Centre, Nottingham, UK*

We present the case of an 11-year-old pre-pubertal Caucasian male who presented with unusual signs of systemic lupus erythematosus (SLE). The initial presentation was with mouth ulcers and neutropenia, treated with granulocyte colony-stimulating factor for 2½ years. Following this a thrombotic, malar, facial rash affecting both cheeks and ears emerged. Investigations at this time revealed positive antiphospholipid and cardiolipin antibodies and weakly positive double-stranded DNA antibodies with low complement, fulfilling the criteria for a diagnosis of SLE. He was initially treated with pulsed methylprednisolone, and then methotrexate, aspirin, hydroxychloroquine, and mycophenolate mofetil and the granulocyte colony-stimulating factor withdrawn. He continues to slowly improve but still has a neutrophil count below normal with occasional mouth ulcers. This is an unusual presentation of SLE due to the thrombotic malar rash but highlights well how the diagnostic criteria for the diagnosis of SLE gradually emerge over time and that individuals with idiopathic neutropenia require screening overtime as they may develop SLE.

G204 CHILDHOOD SARCOIDOSIS: A RARE PRESENTATION OF ADVANCED PULMONARY FIBROSIS IN A YOUNG CHILD

A Kelly, D Eleftheriou, A-M McMahon, C Pilkington. *Great Ormond Street Hospital for Children NHS Trust, London, UK*

Aim: We present the case of an 11-year-old girl with sarcoidosis who presented with uveitis, annular sarcoid skin lesions and advanced pulmonary fibrosis.
Methods: We highlight the features of this rare condition with particular reference to lung fibrosis in this interesting case.
Results: An 11-year-old girl was referred from ophthalmology with bilateral uveitis for 2 weeks. She had lost 6 kg of weight over 1 year while feeling non-specifically unwell. She had shortness of breath on exertion. A florid skin rash was present all over her body with well circumscribed large annular lesions. She had received antifungal treatment without improvement and skin scrapings were negative. Joint examination was normal. There were fine crepitations over both lower lung fields. Chest radiograph showed bilateral basal infiltrates and pulmonary function tests (PFTs) were restrictive in pattern with TLCO 60%. High resolution chest CT demonstrated lung fibrosis. Mantoux test, sputum AFB and culture were negative. She had a normal full blood count and biochemistry, ANA 1:80, rheumatoid factor 70, ESR 28, CRP 8 and an elevated ACE 149. Skin biopsy showed that throughout the dermis there were numerous well-defined epithelioid and giant cell granulomas, some showing slight necrosis. The features were those of granulomatous inflammation. Sarcoidosis was diagnosed. She responded well to oral prednisolone and methotrexate with resolution of the rash and the shortness of breath. Compliance was poor and having not taken methotrexate for 1 month she represented with skin lesions and wrist synovitis. Two years after her initial diagnosis CT of the chest showed increased bilateral lower lobe fibrosis and PFTs showed TLCO 48%. Medications used included prednisolone, methotrexate and azathioprine. Pulmonary fibrosis is recognised as a rare result of childhood sarcoidosis but usually in the older children. Hoffman *et al*[1] described a series of 48 Danish children with sarcoid and none of the patients had evidence of lung fibrosis. Further treatments for childhood sarcoid with lung involvement are mainly based on single case reports or extrapolation from adult data with a lack of evidence for any single agent.
Conclusions: This interesting case highlights the rare presentation of childhood sarcoid with pulmonary fibrosis in a young child. Lack of evidence of further treatment is discussed.

1. **Hoffman AL,** *et al*. *Acta Paediatr* 2007;**93**:30–6.

G205 LINEAR MORPHOEA EN COUP DE SABRE AND PARTIAL SEIZURES: WHAT IS THE LINK?

C Gholam, P Prabhakar, M Glover. *Great Ormond Street Hospital, London, UK*

A previously healthy girl began to experience intermittent abnormal sensations in the left arm at the age of 6 years, with associated abnormal movements appearing a few months later. EEG was normal apart from rhythmic discharges over the right central and Sylvian region during a typical episode provoked by hyperventilation, indicating possible vascular origin. Brain MRI was normal. A year before the onset of neurological symptoms she had developed a discoloured area on the central forehead. By the time she presented to the neurology team she had a lesion typical of linear morphoea (en coup de sabre). Thermal images showed increased temperature in the region of the lesion, with evidence of significantly increased blood flow on laser Doppler. Antinuclear antibody was positive at a titre of 1 in 320. She was treated with pulsed methylprednisolone followed by oral prednisolone and methotrexate. This led to improvement in the skin, but the seizures persisted, requiring treatment with carbamazepine and sodium valproate. The pathogenesis of linear morphoea is not understood. Most of the proposed mechanisms relate to endothelial cell damage leading to increased fibroblast activity. Whether similar processes may be affecting the brain is unknown, although perivascular lymphocytic inflammation, intimal thickening of vessels and intravascular thrombi have been reported on brain biopsy.[1] The case highlights the underrecognised relationship between linear morphoea en coup de sabre and neurological complications, which may persist after the skin changes, have stabilised.

1. **Holland KE,** Steffes B, Nocton JJ, *et al*. Linear scleroderma en coup de sabre with associated neurologic abnormalities. *Pediatrics* 2006;**117**:e132–6.

G206 INCIDENCE OF CHILDHOOD SCLERODERMA IN THE UK AND IRELAND

[1]A Herrick, [1]H Ennis, [1]A Silman, [2]E Baildam. *[1]Arc Epidemiology Unit, University of Manchester, Manchester, UK; [2]Alder Hey Children's NHS Foundation Trust, Liverpool, UK*

Background: Childhood scleroderma encompasses a rare and poorly understood spectrum of conditions and there have been no studies on incidence and prevalence in the UK and Ireland. This study was undertaken to ascertain the incidence of childhood scleroderma in its different forms and to describe the age, sex and ethnicity of affected children.
Methods: The members of five professional associations were contacted: the RCPCH and the Faculty of Paediatrics of the RCPI via the British Paediatric Surveillance Unit (BPSU), the British Association of Dermatologists, the British Society for Paediatric and Adolescent Rheumatology and the UK Scleroderma Study Group. Members were asked to report all cases of abnormal skin thickening suspected to be linear scleroderma or systemic sclerosis (SSc) in children under the age of 16 years first seen between July 2005 and July 2007 (inclusive). Notifying clinicians were sent questionnaires and confirmed cases were followed up 12 months after notification. The denominator population used to calculate incidence rates was derived from ONS (Office of National Statistics) and CSO (Central Statistics Office Ireland) mid-year estimates for children aged 16 years or under for 2005 and 2006.
Results: A total of 185 cases were notified over 25 months. Most were notified via the ''orange card'' scheme of the BPSU and 94 valid cases were confirmed, of which 87 were cases of localised scleroderma (93%) and seven were SSc (7%). This gave an incidence rate of 3.25 per million children per year for localised scleroderma, and 0.26 per million per year with SSc. The majority of localised cases were linear scleroderma: 48 of 87 (68%) children. Of the 87 localised cases, 32 (37%) were reported to have lesions affecting the face and head. 62 cases (66%) were female. 77 (81%) of cases were classed as ''White British''. The mean age at onset was 8.6 years, and the mean time between onset and diagnosis was 1.6 years. Of the 82 cases for whom 12-month data were available, 55 (67%) were reported to be improved.
Conclusions: Linear scleroderma, the form of localised scleroderma associated with the highest morbidity in children, is rare. Childhood SSc is very rare. However, both conditions are associated with severe morbidity and (in the case of SSc) mortality. Current delays in diagnosis may adversely affect treatment outcome.