Pathology

G159 DISCREPANCY BETWEEN NEUROIMAGING AND POSTMORTEM FINDINGS IN A CASE OF GLUTARIC ACIDURIA TYPE I

N. Balgobin[1], M. Farrell[2], S. Ryan[1], E. R. Naughten[1], D. Devaney[1]. [1]*The Children's University Hospital, Temple Street, Dublin, Ireland;* [2]*Beaumont Hospital, Dublin, Ireland*

Introduction: Glutaric aciduria type I (GA1) is an inherited disorder of organic acid metabolism due to a deficiency of glutaryl-CoA dehydrogenase, which leads to an error in the degradation of amino acids lysine, hydroxylysine, and tryptophan. As a result, glutaric acid accumulates in physiological body fluids and tissues and can damage the brain.

Case report: This report describes a 4 year old girl with GA1 and secondary dyskinetic cerebral palsy. Initial presentation was at 18 months with acute encephalopathy post-*Cryptosporidium enteritis*. Management included a protein restricted diet together with aggressive emergency treatment of catabolic conditions that may exacerbate the disorder. A baseline brain MRI showed typical appearances of GA1, and considerable improvement was seen on a repeated study over 2 years later. The clinical course contained recurrent difficulty with intravenous long lines. After the removal of a left subclavian line, she developed acute respiratory distress with opacification of lung fields on chest *x* ray and subsequently died. Postmortem revealed bilateral lung collapse secondary to accumulation of chylous fluid in the pleural spaces and mediastinum caused by tearing of the thoracic duct inlet where it joined a calcified thrombosed innominate vein. Brain histology showed hypoxic changes and established features of GA1 despite clinical and radiological improvement.

Conclusion: This case is unusual in that it demonstrates the clinical and radiological improvement in a child with glutaric aciduria type I (GA1) and shows established histological features of GA1 within the postmortem brain despite these changes. It also highlights the difficulties in management with long term intravenous lines and the importance of the postmortem in identifying unusual complications occurring in these settings.

G160 PERINATAL DEATHS IN THE UK: A BETTER CLASSIFICATION SYSTEM NEEDED?

A. M. Weindling[1], R. Shukla[2], M. Ashworth[2], G. Kokai[2], K. Fleming[3]. [1]*School of Reproductive and Developmental Medicine, University of Liverpool, Liverpool, UK;* [2]*Department of Histopathology, Royal Liverpool Children's NHS Trust, Liverpool, UK;* [3]*Confidential Enquiry into Maternal and Child Health, London, UK*

Background and Aim: In 1995, 40% of perinatal deaths were unexplained[1] and in the following eight year period there has been no improvement in classification with the latest published national figures showing that 45% of perinatal deaths were unexplained in 2002.[2] The purpose of this study was to see whether this situation can be improved by the expert scrutiny of each report individually.

Method: At a single major perinatal centre, all perinatal autopsies of singletons >22 weeks done over a five year period (2000–04) were reviewed by two observers separately (RS, AMW). Terminations because of malformations were excluded. Standard classification systems (Wigglesworth, Fetal & Neonatal, and Aberdeen) were used. In cases where the cause remained unexplained, evidence of fetal growth restriction (weight $<10^{th}$ centile) and taking account of the clinical history (clinico-pathological correlation) were considered.

Results: 271 autopsies (177 stillbirths and 94 neonatal deaths) were reviewed. The cause of death was unexplained after 40% of autopsies, reflecting what was seen in the national data. There was more likely to be a lack of explanation after a stillbirth than a neonatal death (66% *v* 19%, $p<0.0001$). However, when additional information was used (evidence of fetal growth restriction and clinico-pathological correlation), it was possible to provide a probable causal mechanism for death after 74% of autopsies. Nevertheless, while there was failure to identify a significant identified associated factor in only 11% of neonatal deaths, 34% of stillbirths remained without such an identified factor.

Conclusion: Current classification systems do not seem to be able to reflect the complexity of causation of stillbirths. Inclusion of fetal growth restriction/clinico-pathological correlation as an associated factor could enhance current understanding of perinatal mortality statistics. However, existing classification systems could be improved and further research into the aetiology of stillbirths is required.

1. Fourth Annual Report, CESDI 1997.
2. Stillbirth, neonatal and post neonatal mortality 2000–2002. England: Wales and Northern Ireland, www.cemach.org.uk.

G161 AN UNUSUAL PRESENTATION OF LYMPHATIC DYSPLASIA

S. K. Sandhu, G. P. Sinha. *Walsall Manor Hospital NHS Trust, West Midlands, UK*

Background: Lymphatic dysplasia is a developmental disorder of the lymphatic system that classically presents with lymphoedema, an accumulation of lymphatic fluid in the interstitial spaces. We report an unusual presentation of lymphatic dysplasia.

Report: A 3 year old girl with a 4 day history of fever, cough, and difficulty in breathing was admitted. Initially she looked unwell, was tachypnoeic, and clinically there was a suspicion of a left sided pleural effusion. A chest radiograph was performed, which revealed the presence of bilateral pleural effusions. When sampled, a cream aspirate rich in chylomicrons and lymphocytes was demonstrated. A diagnosis of bilateral chylothoraces was made. Treatment involved bilateral pleural drainage and dietary fat restriction. No cause was found to account for the chylothoraces until 4 years later when the child was diagnosed with autosomal recessive lymphatic dysplasia after multiple presentations with lymphoedema.

Conclusion: Chylothorax refers to the presence of chyle, chylomicron-rich lymphatic fluid, in the pleural space. This arises secondary to lymphatic dysplasia of the thoracic duct or one of its main tributaries. Another rare presentation of lymphatic dysplasia is chylous ascites, the accumulation of chyle within the peritoneal cavity. This results from the disruption of the abdominal lymphatics.