Dermatology

G189 ARE CHILDREN WITH RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA OF LOW BIRTH WEIGHT?

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Aim: To investigate the hypothesis that children with Recessive Dystrophic Epidermolysis Bullosa (RDEB) are of lower birth weight than unaffected children.

Methods: Questionnaire based case-control study of 74 patients of the Hospital for Sick Children, Great Ormond Street with RDEB. Birth weight and factors that may influence it were compared to a control group—their nearest unaffected siblings.

Results: Data were obtained on 67 children with RDEB (90.5% response) and 49 unaffected siblings. There was a mean difference in birthweight of 268.7g between RDEB patients and controls, with controls being significantly heavier than RDEB patients. 30% of those with RDEB were Small for Gestational Age in comparison to 10.4% of controls (odds ratio =6.5 p=0.014) and 14% of RDEB patients had severe growth retardation (below 3rd percentile) in comparison to 6% of controls.

A conditional logistic regression was carried out to control for the possible confounding effects of parity, maternal age and maternal smoking between cases and matched controls. The significance of being Small for Gestational Age remained unchanged and was the only significant variable in the model.

Sub group analysis of children with marked skin loss at birth was hampered by small sample size, although 35% of this subgroup were Small for Gestational Age.

Conclusions: Children with RDEB are of significantly lower birth weight than unaffected children. The compromise in growth seen throughout life in RDEB appears to begin in utero. Further work is required to establish the role of skin loss present at birth.

G190 SOMATIC MOSAICISM FOR INCONTINENTIA PIGMENTI IN A NORMAL KARYOTYPE MALE INFANT

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Incontinentia Pigmenti (IP) is a rare dermatogenesis characterised by cutaneous, dental, ocular and neurological abnormalities. This condition is seen almost exclusively in females, as it is segregates as an X-linked dominant trait and is usually lethal in males.

The disease has been linked to Xq28 and approximately 80% of patients with IP have a deletion of exon 4 through to exon 10 of the NEMO gene. This gene is central to many immune, inflammatory and apoptotic pathways. Few affected males have been previously reported, however the majority of these individuals have a 47 XXY karyotype.

We present a case of IP in a healthy male infant with a normal karyotype. He was born at term, appropriate for gestational age, of Romanian parents with no relevant family history. He presented at day 3 of life with a blistering lesion on the medial aspect of his right leg. Extensive viral evaluation was normal. He was referred at 4 weeks of age with recurrence of the rash, again blistering, but now clearly demarcated in a hyper-pigmented linear pattern down the medial aspect of the right leg in the lines of Blascho. The clinical diagnosis of IP was confirmed by PCR analysis of DNA from a skin biopsy sample revealing a deletion of the NEMO gene on Xq28.

The exceptional nature of this case is that the male infant has not only survived but also remains neurologically normal. This is most likely due to the fact that he exhibits mosaicism for the NEMO gene deletion in his somatic cell lines, a finding reported in only a small number of males.

G191 PYLORIC ATRESIA AND NEPHROPATHY WITH EPIDERMOLYSIS BULLOSA: DIAGNOSTIC CLUES FROM DERMATOLOGY

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We report a 2 month-old girl born at 37 weeks gestation, by caesarean section, to non-consanguinous Caucasian parents. Pyloric atresia was diagnosed on an antenatal scan and karyotyping was normal. Fragile skin and blisters were noticed soon after birth. Laparotomy on day 4 revealed a large stomach, thickened pylorus and a complete pre-pyloric membrane. She underwent pylorotomy, excision of the pre-pyloric membrane and Heinele-Miculicz pyloroplasty and recovered without complications.

A clinical diagnosis of Junctional Epidermolysis Bullosa with Pyloric Atresia (JEB-PA) was confirmed by skin biopsy, which showed a split in the dermo-epidermal junction (DEJ) through the lamina lucida. Indirect immunoflorescence demonstrated normal bright staining along the DEJ using the following antibodies: GB3 (laminin 5), LH 7.2 (Type VII collagen), HD 121 (plectin) excluding Herlitz JEB, dystrophic EB and EB simplex with mucosal dystrophy. There was reduced staining with 439-9B (α4 integrin) and GOH 3 (β6 integrin).

Post operatively, she has suffered occasional blistering of her skin and mucous membrane, but is otherwise thriving. Hair and nails are normal.

Discussion: The association of JEB and PA is well known and is due to mutations in the integrin α6 or β4 gene (IGTA6 or ITGB4). Prognosis is variable. Most cases are serious and often lethal in early infancy similar to the Herlitz form of JEB but several patients with a milder phenotype have been reported. Survivors tend to develop obstructive uropathy and renal function must be monitored. Our patient has a bifid left kidney with a mild prominence of her left renal pelvis demonstrated on ultrasound examination, and normal renal function.

The molecular cause of this condition, established by the study of the skin, should be of interest to gastroenterologists and nephrologists.