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 being Small for Gestational Age. Given that gestational age was adjusted for age and smoking between cases and matched controls, the significance of birthweight of 268.7g 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 Smaller 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.

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 blistersing 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 blistersing, 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.