on newborn hearing screening and should check if screening has been completed or that babies are referred for audiological assessment.

Child Protection Special Interest Group

717 EXPLORING THE RADIOLOGICAL COMPLEXITY OF ABUSE IN CHILDREN UNDERGOING SKELETAL SURVEYS – A PICTORIAL REVIEW

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Background Studies have reported an increased incidence of physical abuse in children during the COVID-19 pandemic, likely a result of an interplay of factors, including social isolation, lack of support systems and financial uncertainty, emphasising the importance of this complex issue.

When abuse is suspected, a cascade of investigations is triggered, including radiological imaging, performed in accordance with national guidelines. For children under two, this includes skeletal survey (and computed tomography (CT) head scan if under one or in the presence of clinical evidence of head trauma). Skeletal survey is performed on a case-by-case basis for older children.

Interpretation of findings requires an understanding of typical patterns of skeletal injury caused by physical abuse, as well as recognition of skeletal indicators of neglect and radiographic mimics of abuse.

Objectives The objective of this review is to describe the spectrum of radiological findings in the investigation of suspected physical abuse. Findings on skeletal survey are presented for a group of children, chosen to include:

1. Typical patterns of skeletal injury
2. Manifestations of neglect
3. Mimics of abuse

For each case, presenting circumstances and outcome are also described, in order to tell a series of important stories.

Methods Skeletal surveys, performed for the investigation of suspected abuse, over a five-year period were reviewed. Of those with confirmed skeletal abnormality, several were selected for presentation.

Results Over a five-year period, eighty-nine skeletal surveys were performed for the investigation of suspected abuse. Skeletal abnormality was identified in thirty-one cases.

Cases chosen to present include children aged between 4 weeks and 2 years, from various social backgrounds. Investigations were prompted by various presenting circumstances, including poor feeding/vomiting, limb injury with suspicious mechanism, unresponsive episodes and direct allegations of physical abuse. In one case, suspicion of abuse was raised when rib fractures were found incidentally on a chest radiograph.

Cases are described in which there are typical patterns of inflicted fractures, including cases of multiple fractures.

Cases in which there are skeletal manifestations of neglect are described, including severe metabolic bone disease.

Cases of skeletal abnormality, concluded to be non-pathological (e.g. normal variants and physiological changes) are described

The role of follow-up imaging is described.

In terms of outcome, some children were returned to the custody of their parents or family, with comprehensive Child Protection Plans in place while, for others, emergency adoption or Foster care placement was required.

Conclusions While it is difficult to confidently describe a 'typical' case of physical abuse, there are recognised clinical presentations, as well as patterns of skeletal abnormality and associated imaging findings to support a diagnosis of abuse in children. Follow-up imaging is important, particularly when findings are inconclusive or there is suspicion of occult injury. Fundamental to comprehensive assessment of imaging is a detailed understanding of recognised radiographic mimics of abuse.

While focusing on radiological findings, this review also serves to highlight the profound consequences of abuse in children, which extend beyond the physical. In addition, it emphasises the complexity of child abuse which can present in children of any age, from any social background.

British Paediatric Neurology Association

718 A *DYNC1H1* MUTATION ASSOCIATED WITH SPINAL MUSCULAR ATROPHY WITH LOWER EXTREMITY PREDOMINANCE (SMA-LED) AND MIXED UPPER AND LOWER MOTOR NEURONE FEATURES

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Background Spinal muscular atrophy with lower extremity predominance (or SMA-LED) is a group of rare, autosomal dominant disorders affecting lower motor neurones and characterised by weakness and atrophy of the lower limb muscles. Variants in two genes have been linked with SMA-LED: *DYNC1H1* and *BICD2*.

Objectives To present the first familial case series of a mixed upper and lower motor neurone clinical phenotype of SMA-LED, associated with a mutation in *DYNC1H1*.

Methods Case series of three family members (father and two sons) across two generations.

Results We describe a family with a unique, mixed upper and lower motor neurone clinical phenotype of early onset SMA-LED, inherited in an autosomal dominant manner, caused by a missense variant c.1808A>T (p.Glu603Val) in the cytoplasmic dynein 1 heavy chain 1 gene (*DYNC1H1*). The father had been believed to have a purely orthopaedic pathology (talipes), and the eldest son was not diagnosed until his second year of life.

Conclusions Based on ACMG variant classification guidelines, we suggest that this variant in *DYNC1H1* be reclassified as 'Likely Pathogenic'.