Osteoporosis / osteopaenia in recessive dystrophic epidermolysis bullosa (RDEB)

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At GOS, we have recently identified several RDEB children with moderate to severe bone pain, both with and without radiological evidence of micro-fractures. Pain, when present, is usually located in the spine, feet and knees. Bone mineral density (BMD) scans reveal abnormally low levels, although it seems that interpretation of these scans in children is not straightforward. Laboratory investigations demonstrate some slightly abnormal calcium and phosphate levels, but not in all patients, or consistently in the same patients. Vitamin D levels have been infrequently measured, but available results are normal. A literature search for similar cases found only one reference, (1) the patient reported being an adult sufferer.

All the children are severely affected by their EB in terms of:

- limited oral nutrition (largely as a result of microstomia and oro-pharyngeal and oesophageal strictures)
- heavy reliance on gastrostomy feeds for maintenance of nutrition
- extent of poorly-healing, or non-healing skin lesions
- limited mobility, despite pro-active physiotherapy (some are wheelchair-bound)

A number of factors were initially considered as being potentially responsible:

- inadequate intake of dietary calcium / phosphate / Vitamin D
- insufficient exposure of skin to sunlight
- insufficient weight-bearing exercise
- steroid administration
Points to consider

- All the gastrostomy feeds are theoretically more than ample in terms of calcium and phosphate, as compared to the Reference Nutrient Intake (RNI) (2) for age and sex-matched unaffected children, supplying 120 - 245% RNI for calcium and 120 – 183% RNI for phosphate. In terms of vitamin D, the feeds provide at least 105% of the amount considered advisable in United Kingdom for "at risk individuals" ie. 7mcg/day for children and 10mcg/day for adolescents.

- These children tend to spend little time in the sun and varying areas of skin are covered by dressings.

- There may be an association between :- skin lesions present at birth, or developing shortly after (especially those on the feet), reluctance to weight-bear despite analgesia (e.g. Amitriptyline) and the subsequent development of bone pain. If not actually wheelchair-bound, most of these children are extremely limited in terms of weight-bearing exercise which would normally stress the joints and bones.

- Steroid administration is kept to a minimum, but can be unavoidable as part of the skin care strategy.

Our current management

Routine measurement of BMD is being undertaken in all severely affected children. Calcichew D₃ Forte (500mg Ca and 10mcg [400units] cholecalciferol) is prescribed for most of the children with pain and / or radiological changes, even if the diet is theoretically adequate. Some children receive intravenous infusions of Pamidronate (a bisphosphonate which reduces the rate of bone turnover).

The EB team would like to organise a randomised, controlled trial in order to try to elucidate more data about osteopaenia / osteoporosis in EB. Pain control management is a priority.

Tentative conclusions, but watch this space!

Although it seems very likely that the cause of the bone abnormalities seen in our small group of patients is multi-factorial, the main "culprit" is likely to be immobility. Of course, dietitians should always monitor feeds for nutritional adequacy. However, this should be done whilst bearing in mind factors which are specific to the condition, such as low sunlight exposure. It should not be assumed that because the theoretical requirement is being met, the intake is adequate.
As always, the importance of the multi-disciplinary team approach cannot be over-emphasised, so if a dietitian feels that their patient’s intake should be supplemented, the situation should be discussed with fellow EB team members and a holistic approach taken.
