Craniotabes: a case of congenital deformation of the skull

This article presents a striking cranial deformity in a newborn baby in the context of an abnormal lie *in utero* compounded by biochemical vitamin D deficiency in the mother and baby in a high risk ethnic group. This case highlights the importance of prophylactically addressing adequate vitamin D nutrition in pregnancy to prevent occult bone disease in newborns.

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Keywords

neonate; vitamin D deficiency; craniotabes; newborn examination; cranial deformity

Key points

Parmenter KF, Lok A., Miall L. Craniotabes: a case of congenital deformation of the skull. *Infant* 2019; 15(4): 155-56.

1. Maternal vitamin D deficiency remains prevalent, especially in Asian and African ethnic groups.
2. Early recognition and treatment of vitamin D deficient mothers and their babies is important.
3. This case highlights the significance of antenatal screening and prophylactic vitamin D supplementation for mothers at high risk of vitamin D deficiency.

The case

A newborn baby girl presented at term delivery with a strikingly flat indentation on the right parietal aspect of her skull and bilaterally swollen eyes after an emergency caesarean section due to a difficult brow occipito-transverse lie following an uncomplicated pregnancy (**FIGURE 1**). An X-ray confirmed a depressed skull fracture with soft tissue swelling (**FIGURE 2**). A cranial ultrasound was unremarkable. No deformity had been suggested on antenatal scans.

The baby had a vitamin D level of <20nmol/L (normal range 75-250nmol/L) with an otherwise normal bone profile. Her calcium, alkaline phosphatase and phosphate levels were within the normal biochemical limits. The mother, who was of Asian background, also had vitamin D insufficiency with a level of 37nmol/L. There were no other signs of bone disease and the infant was diagnosed with craniotabes secondary to *in utero* vitamin D deficiency exacerbated by malposition. She was commenced on alphacalcidol with follow-up in the community outpatient clinic. At six months of age, her development was normal. The deformity was still present but less visible and no long-term co-morbidity was expected. Her vitamin D level was 34nmol/L.

FIGURE 1 Craniofacial appearance showing the flat indentation of the skull.

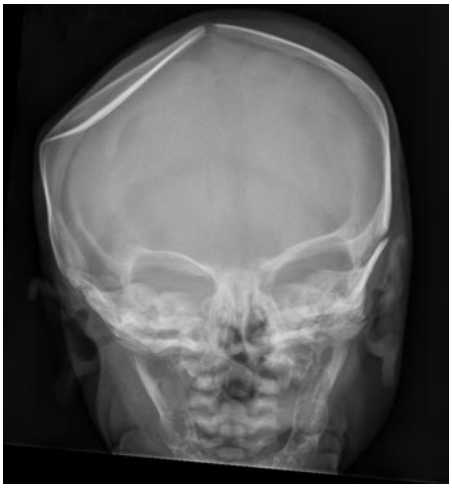

FIGURE 2 X-ray delineation of the skull deformity.

Discussion

Craniotabes

Craniotabes is a softening of the skull bones described as the presence of soft bones with inward collapse when pressure is applied by the examiner's fingers, which typically snap back when the pressure is relieved – the 'ping-pong ball' effect. Craniotabes is common in infants, particularly those born premature, occurring in up to one third of all newborns. It has been suggested that craniotabes in otherwise normal neonates is associated with vitamin D deficiency *in utero*. It is harmless in the newborn infant unless it is associated with other co-morbidities, which include osteogenesis imperfecta, hypervitaminosis A and

congenital syphilis.[2]

In this case, early descent of the head in a vertex position may have elicited prolonged forceful pressure thus diminishing cranial mineralisation and affecting the superior portions of the parietal bones. Compressive effects can occur in up to 30% of newborn babies.[2-4]

Vitamin D

Vitamin D is a fat-soluble vitamin that is responsible for the maintenance of calcium homeostasis within the body. Its two main forms are D_2 (ergocalciferol) and D_3 (cholecalciferol), the latter being produced in the skin during exposure to sunlight. Both ergocalciferol and cholecalciferol are metabolised by the liver to make the major circulating forms of the vitamin 25-hydroxyvitamin D_2 and 25-hydroxyvitamin D_3. The kidneys play a vital role in turning the circulating forms of the vitamin into 1,25-dihydroxyvitamin D_2 and 1,25-dihydroxyvitamin D_3, the active forms of vitamin D that then go on to help increase absorption of calcium and phosphate from the small intestine, aid in the mineralisation of the bone matrix and inhibit the synthesis of parathyroid hormone.

Vitamin D can be obtained from the diet and is synthesised in the skin by exposure to sunlight, however inadequate vitamin D is common and high-risk groups for vitamin D deficiency include:

- pregnant and breastfeeding women
- infants of twin and multiple pregnancies
- adolescents
- obese individuals
- those with reduced sun exposure, eg living in the northern latitude, Asian and African descent (dark skin needs more sunlight), wearing concealing clothing, immobility, excessive use of sun block.

The British Paediatric and Adolescent Bone group defines vitamin D deficiency as a serum level of vitamin D below 25nmol/L, and insufficiency as between 25 and 50nmol/L. It is known that clinically, insufficiency can cause muscle aches and pains and deficiency can cause seizures, cardiomyopathy, myopathy, rickets and poor growth.[5] Routine testing of vitamin D levels in infants is not recommended but a test is recommended when an infant or child is symptomatic and has risk factors.

Yew et al conducted an audit of infants who were tested for vitamin D levels. Of the 81 infants in the study, there were 45 infants categorised as low vitamin D (<50nmol/L).[6] Over two thirds of these infants were from an Asian or African background. Although none of these were described as having an abnormal head size or shape, 24 were described as having a large fontanelle/craniotabes.

Management

For craniotabes, the prognosis is excellent with mean resolution of appearance within two to three months.[2,7] For infants with identified vitamin D deficiency, treatment with supplements should focus on functional rather than biochemical outcomes. The Department of Health and Social Care[8] and the Royal College of Paediatrics and Child Health[9] recommend that infants should receive a daily vitamin D supplement of 400IU from birth until one year of age, unless consuming more than 500mL of formula milk a day, as infant formula is routinely fortified with vitamin D. Children aged one to four years of age should receive a daily supplement of 400IU. Children with darker skin should take between 400-1000IU vitamin D_3 up to 18 years of age, to ensure a deficiency does not occur.

As with most medical conditions, prevention is better than cure. Pregnant and breastfeeding mothers should be advised to ensure a diet high in calcium and vitamin D diet and/or vitamin D supplements along with suitable time in the sun. We would like to highlight the importance of antenatal screening of vitamin D in high-risk populations and the treatment of infants and mothers at risk following detection of deficiency.

Acknowledgement

Dr Parmenter and Dr Lok would like to thank Dr Miall for his time and expert comments when reviewing the content of this case report.

Parental consent

The authors received written consent to publish this report from the patient's parents.

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