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COMMENTARY

The death of temporary brittle bone disease is premature

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> Where all men think alike, no one thinks very much
> (Walter Lippmann)

In this issue of Acta Paediatrica is an article on temporary brittle bone disease (TBBD) by Paterson (1). Paterson will be forever linked to TBBD as he coined the term in the early 1990s to describe a group of infants who had multiple unexplained fractures (MUF) in the first year of life (most under 6 months of age). He thought TBBD was a transient, intrinsic bone disorder as the fractures were limited to the first year of life, and that it was highly unlikely that these infants were abused as there was no bruising (2,3). However, these infants had X-ray fracture characteristics that are thought to be pathognomonic of child abuse including: (i) metaphyseal fractures; (ii) posterior rib fractures and (iii) fractures at various stages of healing. Paediatric radiologists had long preached that these features indicated child abuse and paediatricians accepted this dogma without question. Paterson's proffer of TBBD as a mimic of child abuse was a direct challenge and threat to the child advocacy establishment (CAE) who strongly disagreed with Paterson's assertion that this was an intrinsic bone disorder, as Paterson could not provide evidence at that time of a specific bone disease or predisposing factor(s) that might explain TBBD.

Paterson testified in the legal proceedings of infants with MUF in which he diagnosed TBBD, but the CAE diagnosed child abuse and the children were removed from their parents. In some of these cases, the infants were returned to their parents, because of Dr Paterson's testimony that defended the concept of TBBD in the court room. Dr Paterson's success in returning children to their parents, in the face of a diagnosis of child abuse by the CAE, angered the CAE (4). Because the CAE was having a difficult time prevailing in attacking his message in the court room, the CAE decided to attack the messenger, Dr Paterson, in the court room and brought successful proceedings of disbarment against him in 2004 by the GMC in England leading to Dr Paterson's apparent premature retirement from medicine (5).

By having Dr Paterson disbarred and publishing policy statements and reviews in publisher friendly journals which explicitly state that TBBD does not exist and is a ruse for child abuse, the CAE had hoped to forever bury TBBD (6,7). After all, accepting the existence of TBBD would be tantamount to the admission of the incorrect diagnosis of child abuse in thousands of cases of infants with MUF over the past 30 years. Much is at stake for both the families enmeshed in this issue and for the physicians who diagnose child abuse in these cases.

Invited Commentary to Colin Paterson. Temporary brittle Bone Disease: Fractures in Medical Care.

OBSERVATIONS THAT INDICATE TBBD IS NOT CHILD ABUSE

Thus, Paterson's study in this month's Acta Paediatrica reopens the issue of TBBD and takes on much significance. The observation in this study that the fractures of TBBD can occur while the infant is in the care of medical providers is one more piece of evidence that TBBD is not a ruse for child abuse. Other observations that TBBD is not child abuse include:

- *The absence of significant bruising*. Because the force required to cause the failure of skin integrity (bruise) is far smaller than the force needed to cause the failure of bone integrity (fracture), one would expect significant bruising in infants with TBBD if indeed it was battering that caused the fractures (8,9). Absence of bruising in these infants, some who have as many as 25 fractures, begs for an alternative medical explanation.

- *Multiple rib fractures without any severe internal thoracic injury*. Garcia et al. (10) noted that in children who have four or more rib fractures from trauma (including child abuse), there was always severe internal thoracic injury such as pneumothorax, haemothorax or lung contusion. The absence of severe, internal thoracic injury in infants with TBBD who have four or more rib fractures suggests that the forces that caused the rib fracture were minimal, and thus there an intrinsic bone disorder is far more likely than child abuse.
- *The frequency distribution of the age of presentation of infants with TBBD*. Infants with TBBD have a peak age of presentation at about 2 months of age and few infants with TBBD present after 6 months of age (11). If, indeed, TBBD is a ruse for child abuse one would expect to see many infants present at later ages. However, such is not the case. The early age of presentation with relative close proximity to birth suggests the clue to understanding TBBD lies in understanding those factors in the prenatal and perinatal time period that might transiently influence bone strength in young infants.
- *Paterson's follow-up of infants with TBBD who were returned to their parents*. In 65 infants (mean of 4.9 years, range: 0–11 years) whom he diagnosed with TBBD counter to the diagnosis of child abuse by the CAC who were returned to their parents, 47 with conditions, there was no evidence of subsequent child abuse in those infants returned to their parents (4).

  Recently another infant with TBBD has been described who, like those infants in Paterson's article, also incurred multiple fractures while in medical care (12). This 3.4 kg of 36 weeks gestational age was born with a large cervical tumor that was resected at 10 days of age, and the infant remained in the neonatal intensive care unit for some 2 months. Serial chest X-rays unequivocally show that the infant developed multiple, unexplained rib fractures at about 7 weeks of age.

WHY HAS TBBD MISTAKENLY BEEN CALLED CHILD ABUSE

In the 1970s, infants with MUF were called child abuse as a default diagnosis. When parents could not explain why their infant had 20 fractures, the CAE thought this lack of explanation was indicative of child abuse as the CAE did not think the physical findings were consistent with such a history. This conclusion was reinforced by the radiology of child abuse. Paediatric radiologists have made two flawed assumptions in their interpretation of the skeletal films in infants with MUF. First, they have incorrectly assumed that a plain X-ray can judge bone strength – it cannot. Paediatric radiologists will often presume that because the bones of an infant with MUF have a normal whiteness on plain X-ray that the bones are necessarily of normal strength, and thus the fractures in this infant must have been caused by excessive and violent forces, i.e. child abuse. Bone strength is determined by bone density, bone architecture (geometry) and bone quality (13). However, a plain X-ray cannot reliably determine any of these three determinants of bone strength. Regarding bone density, a plain X-ray must lose 30–40% of its bone density before a radiologist can appreciate it (14).

The second flawed assumption is that the three previously noted X-ray findings are pathognomonic of child abuse – they are not. There are many reports of these features described in scenarios other than child abuse including the bone disease of prematurity (15–17). Yet, the plain X-ray often trumps any other information in cases of infants with MUF and is the engine that drives cases to be called child abuse.

WHAT CAUSES TBBD?

If TBBD is a real medical condition, what causes it? In 1994, I began studying infants with MUF using bone density measurements. Most of these infants with MUF had a TBBD phenotype similar to what Patterson had recently described. Based on my experience in evaluating infants with TBBD by clinical history and bone density measurements, I proposed in 1999 that decreased foetal bone loading through decreased foetal movement was one factor that could cause TBBD, a hypothesis that is in accord with the Utah paradigm, the widely accepted paradigm of bone physiology popularized by Frost (17–19). Both bone density and bone architecture are different in infants with TBBD compared with those in normal controls, and these differences lead to a weaker bone strength in infants with TBBD (18,20). Bone ultrasound findings in newborns have supported the concept that foetal bone loading is an important determinant of infant bone strength (21–23).

There are other factors that can cause/contribute to TBBD including vitamin D deficiency, prematurity, gestational diabetes and likely other factors, not yet described (24–27). Thus, a critical analysis of cases of TBBD can provide insight into the determinants of infant bone strength. Several conclusions about infant bone strength can be drawn from this scrutiny:

- Multiple genetic loci and multiple environmental factors determine infant bone strength.
- Many of the environmental factors that influence infant bone strength are variable and act in a transient and variable fashion and include the following: foetal bone loading, vitamin D status, prematurity and gestational diabetes. Infant bone strength is thus a multifactorial characteristic and can vary from infant to infant.
- These transient environmental factors can adversely affect infant bone strength and present as TBBD. TBBD can result from the negative influence on bone strength of only one of these environmental factors acting singularly or from several of these environmental factors acting additively.
- A plain X-ray is inadequate to judge infant bone strength.

CONCLUSION

The CAE plays a critical role in protecting children from harm. However, their influence sometimes goes beyond what is reasonable in making a diagnosis of child abuse. Such is the case in the infant with MUF. Dr Paterson has done a great service to society by challenging the scientifically unfounded belief system of the CAE in their approach to the evaluation of the infant with MUF. When all of the scientific evidence is considered, it is highly likely that an entity called TBBD does exist and reflects previously unappreciated factors that influence infant bone strength. The concept of TBBD has escaped appreciation for so many years, because of the zealous adherence to a flawed belief system that incorrectly assumed that a plain X-ray can evaluate bone strength and that there are pathognomonic radiographic features of child abuse in infants with MUF.

The CAE had thought they had done away with TBBD as a real entity and had banished Dr Paterson into retirement. Not so. As evidenced by Paterson's study in this issue of Acta Paediatrica, Dr Paterson's absence from this issue has only been 'temporary', and the observation in this article is one more piece of evidence that supports the existence of TBBD.

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