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Osteogenesis Imperfecta and Non-accidental Injury: Problems in Diagnosis and Management

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Summary

It has been noted in the literature that Osteogenesis Imperfecta is frequently mistaken for non-accidental injury. This article serves to illustrate the difficulty in differentiating between the two conditions and that they can occur concomitantly in one patient.

Key Words: Osteogenesis Imperfecta, Non-accidental injury, Child abuse

Introduction

Non-accidental injury or child abuse is now reported frequently in many developed and developing countries. In most cases, the condition is very obvious both from the history and on physical examination of the children concerned. However, the clinician, in his evaluation of a suspected case of child abuse with fractures, must also consider concomitant medical conditions so as to organise a more comprehensive plan of action and thus avoid unnecessary separation of the child from his/her family. The most frequent of these conditions is Osteogenesis Imperfecta (OI), a hereditary condition occurring in about 1 out of 20,000 of the population[1,2]. Osteogenesis Imperfecta is a heterogenous group of disorders often brought to medical attention because of recurrent and multiple fractures, resulting from biochemical disorders of collagen or collagen production. Sillence, Senn and Danks[1] from their experience of 155 patients in

Table I
Classification of Osteogenesis Imperfecta (Sillence types[1,2])

Sillence	Clinical Features	Inheritance
I	Mild to moderate severity. Little impairment of growth. Blue sclerae at all ages	Autosomal dominant. New mutation occurs frequently
II	Very severe disease causing stillbirth or early neonatal death	Not known in most cases;autosomal recessive in some;new dominant mutation in some
III	Severe disease with antenatal fractures in most cases. Progressive deformity common. Severe impairment of growth Blue sclerae in some but not all cases	Autosomal recessive in most cases
IV	Mild to moderate severity. Impairment of growth may occur. White sclerae in older children and adults. May have blue sclerae in early childhood	Autosomal dominant. New mutation occurs frequently

*Subdivided into A (no dental abnormality) and B (*dentinogenesis imperfecta *present)*

Victoria, Australia suggested 4 distinct types of OI (see Table I). In each of these 4 types of OI, there is a variable expression, even within families, and long periods without fractures are recognised[1,2].

Not unexpectedly, especially in children with the type IV variant, misdiagnosis of child abuse may occur especially if there is no family history (as in fresh mutations) or if no proper family history is obtained[1,3]. A case of a three-and-a half-year-old child and her family, is presented here to illustrate the problems encountered in a child with suspected non-accidental injury and a positive family history of OI.

Report

CSY, a 3 ½-year-old girl, was the youngest child of a family of four, other siblings being 24, 22 and 20 years of age and who were well with no history of fractures. She was born by normal vaginal delivery and was well at birth and for the first 3 years of her life. Her father was a mildly deaf, 67-year-old odd-job man and mother a 44-year-old rubber tapper. Their marriage was non-consanguineous.

Living in the same house were her 20-year-old sister (who had just come back from Singapore one month before CSY's first admission and had set up a hairstyling business in the house) and her sister's boyfriend, both of whom looked after this little girl when the mother was out tapping rubber.

She was first admitted on 21.1.92 to the gynaecological ward of a General Hospital because of per vaginal bleeding allegedly from a forward fall from a chair whilst at home. She was looked after by her sister and her sister's boyfriend at the time the incident occurred. Findings during this admission showed a 1 cm.

hymenal tear from the fourchette along the posterior vaginal wall and a 0.5 cm. tear at the fourchette. This was sutured under General Anaesthesia. In addition, the doctors also noticed multiple bruises over the knees, chin and hand, suprapubic region and right anterior superior iliac crest. The child was referred for further assessment to exclude non-accidental injury but was inadvertently sent home.

The child was next admitted this time to another hospital on 23.3.93 with bilateral periorbital bruising noted on the day of admission, a haematoma over the skull vertex and right parietal area, and a bruised left knee. This was allegedly secondary to a knock on the edge of the car door, four days earlier, as the family was rushing her to see a doctor after a febrile convulsion. The family had only noted her "scalp swelling" when she had periorbital bruising on the day of admission. While in the ward, small fresh bruises on the limbs were seen on 2 occasions.

On two occasions following home leave, the child had numerous bruises over her trunk and limbs on her return. The child's parents, siblings and sister's boyfriend denied any physical abuse and refused admission after home leave on the second occasion. Non-accidental injury was strongly suspected and the Social Welfare Department was duly informed to supervise the child and family closely.

When home visiting by the Welfare Department was made a few weeks later, the child was found to have her left arm splinted and in a sling. The parents had apparently sought medical attention three weeks earlier for pain at the shoulder region (noted initially on waking up) but was reassured and sent home. When the pain persisted, the parents had sought traditional treatment. The parents were instructed by the Social Welfare Officers to bring the child to the local district hospital, where a diagnosis of transverse fracture of the left upper humerus, as well as multiple fractures, noted on skeletal survey (see below) was made. A family history of Osteogenesis Imperfecta was elicited there. She was then referred to the Hospital Kuala Lumpur for diagnostic work-up of OI vs non-accidental injury.

On examination the child was small for her age (weight 12.0 kg.; height 100 cm.). She had normal sclera, with normal dentition and hearing. There was no bony deformity of the left humerus. However there was fixed flexion deformities at the wrist joints and restriction of movements of metacarpophalangeal and proximal interphalangeal joints. Old bruises were found over the right lateral thigh. There was no hyperelasticity of skin or joints. The child had normal sensitivity to pain.

Psychiatric assessment showed the child to be extrovertive with a close relationship to the family, especially her mother. The family and her sister's boyfriend blamed her fractures on the "bone disease" and denied any physical/sexual abuse. Interview of the child did not reveal any conclusive evidence of physical or sexual abuse. It was felt that physical abuse by the mother was extremely unlikely in view of the caring relationship between mother and child, noted by the nursing and medical staff. It was however noted by the staff and neighbouring patients that the sister's boyfriend tended to handle the child roughly.

In the ward, the child developed a small haematoma over the right proximal third interphalangeal joint after rubbing her knuckles on the floor, as well as mild bruising over her thighs and back from mild trauma witnessed by ward staff.

Coagulation profile, platelet function studies and collagen screening were normal. Skeletal survey at the district hospital had showed multiple fractures of varying ages of the left and right clavicle, left humeral shaft, right seventh rib, proximal ends of the second and third metacarpal bones of the right hand, compression fractures from the fourth to the seventh thoracic vertebrae as well as the occipital fracture noted on the previous admission. Skeletal survey at the General Hospital Kuala Lumpur revealed the same fractures, except for those of the metacarpal bones and occipital bone, which were no longer visible radiographically. The bones were reported to be of normal density (see Figures 1a and 1b).

Out of the father's 4 remaining live siblings, all 3 females suffer from OI, whereas the other surviving male is normal (see Figure 2). Two of the affected females aged 55 and 53 have been bedridden for more than

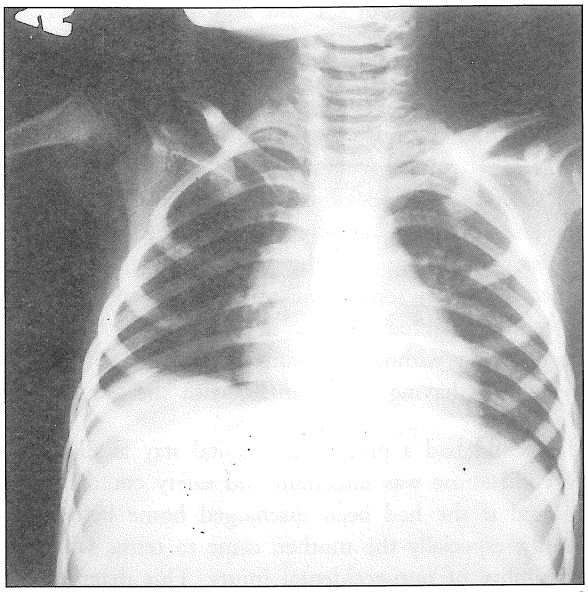

Fig. 1a: Bilateral clavicular fractures and fracture 7th rib posteriorly

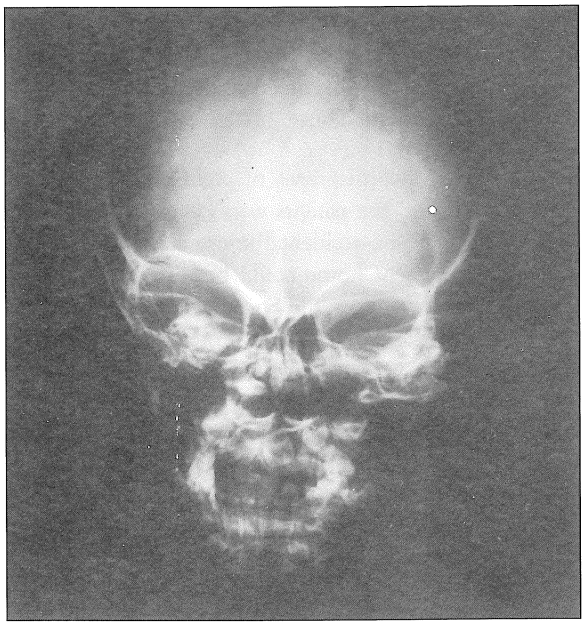

Fig. 1b: Occipital skull fracture

30 years, with multiple non-united fractures. The other, aged 52, was short (height of 137 cm), had multiple bony abnormalities and was only mobile on crutches. All three had normal coloured sclerae and their hearing appeared grossly normal. All three had lost their teeth

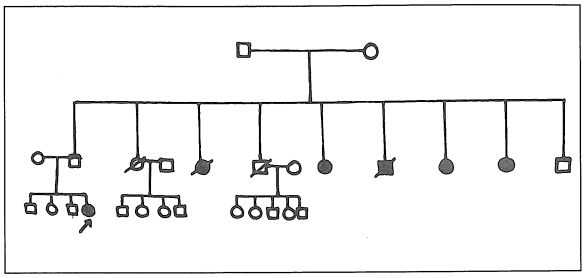

Fig. 2: CSY's family tree showing affected family members. All affected aunts/ uncles have not married because of their disabilities

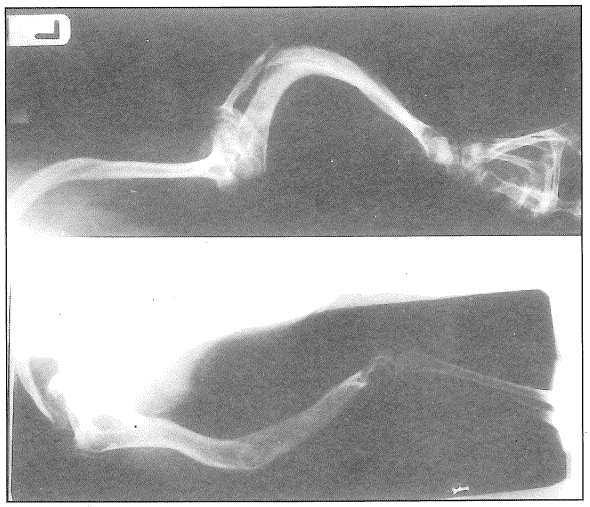

Fig. 3: X-ray of the upper limbs of CSY's aunts showing severe deformity and osteopaenia

in their middle age. X-ray findings of the first two women were consistent with OI and prolonged immobilisation (see Figure 3). Two other siblings (1 female, 1 male), who were short and who had bony abnormalities, had passed away in their thirties. Two unaffected siblings had died from the complications of hypertension.

The girl's father appeared unaffected although he had conductive deafness. He was of normal height and sclera with no past history of fractures, and had normal X-ray findings. He had lost all his teeth 10 years previously but his dentition was normal in his thirties.

In the ward, a few new bruises were detected on the child following minor or undetected trauma. Generalised severe bruising with haematoma formation was noticed only after 2 visits by the sister's boyfriend. This included an adult bite mark on the child's lower limb. However, no new fractures were observed. After informing the Social Welfare Department and the local Police, the child was again discharged home under the care of her mother after more than 2 months in hospital.

After 10 days at home, the child was brought in by the mother for bilateral ear bleeding with purulent discharge noted on that day. This was diagnosed by the Ear, Nose and Throat Specialist as being secondary to otitis externa and otitis media. She was also found to have bilateral fractures of the ulnae/radii, a new occipital fracture (asymptomatic) and fresh bruises on her chest and back. There was no history of trauma according to her mother.

Social Welfare and police officers felt that OI rather than physical abuse was the cause of her problems as reports by neighbours were favourable towards the family. However, circumstances such as will be discussed below made the diagnosis of OI type IV with non-accidental injury more likely.

Discussion

It is not often reported of child abuse occurring in a patient with osteogenesis imperfecta. In our patient, a diagnosis of OI is very probable given the multiplicity of fractures occurring over a period of time after allegedly minimal or no trauma.

The presence of OI in the girl's 4 aunts and 1 uncle helps towards making the same diagnosis in this child. The absence of disease manifestation in the father (other than perhaps his deafness which could be due to other causes) does not preclude the diagnosis of OI as there are families on record with transmission of OI type IV through individuals who appear to be normal with few or no fractures. Furthermore, it has been noted that frequently the only x-ray findings in the milder forms of OI in the early years of life have been the fracture(s) itself, with a normal rate of healing without any evidence of osteopenia[1]. The child's short stature could be explained by both OI and non-accidental injury.

On the other hand it also cannot be denied that the child was physically and possibly even sexually abused. The hymenal and vaginal tear at 6 o'clock with multiple bruises over the body, as stated earlier, suggested possible sexual as well as physical abuse when the child was first admitted. Later incidents of severe bruising occurring and the presence of a bite mark, especially when sent home on leave or when visited by the suspected abuser, as compared to during hospital stay without any visitors, also points to abuse by persons having close contact with the family.

This child had a prolonged hospital stay as diagnosis of child abuse was uncertain and safety could not be assured if she had been discharged home before the family, especially the mother, came to terms with the possibility of non-accidental injury. This diagnosis of concomitant non-accidental injury became obvious when the child was sent home and readmitted on 2 occasions with fractures and bruises. The mother denied that the child had been physically more active at home than in hospital. However, by this time, the mother was more convinced of the possibility of concomitant physical abuse.

Once the definitive diagnosis of child abuse as well as OI was made and the parents were convinced that there could have been non-accidental injury as well, the child's safety on being sent home could be more assured with regular home visiting by the Social Welfare services and counselling of the parents as to protection of the child with regards to OI and physical abuse. At the same time, instead of the child being sent to a Welfare Home, it was recommended to the family that the suspected abuser, the sister's boyfriend, be excluded from living in the same house and not to be left alone with the child at any time. The Social Welfare officer who had become closely involved with the family would continue supervision, together with the local paediatrician and the doctors here. Prosecution of the case was difficult without eyewitness accounts or disclosures by child or family members.

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"Koro"-like Syndrome Affecting the Tongue – A Case Report

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Summary

A 52-year-old man presented with a 2-year history of episodic retraction of his tongue into the throat with a belief that he will die if the retraction is complete. The presentation is similar to koro except that the tongue is involved instead of the penis. It appears that retraction taxon can involve other organs and may not necessarily be culture bound.

Key Words: Koro, Tongue, Retraction taxon

Introduction

A taxon refers to groupings based on similarities without specifying its level of abstraction[1]. This term is used to group the usually rare and so called 'culture bound' syndromes, though nowadays some of the conditions are not entirely regarded as culture bound. Simons has described seven taxa: startle matching