

Shaken Baby Syndrome Suggestive of The Diagnosis of Osteogenesis Imperfecta in Newborn

Ali Annagür¹, Hüseyin Altunhan², Bilge Burçak Annagür³, Sabahattin Ertuğrul⁴, Rahmi Örs⁵

ABSTRACT

Physical child abuse may occur in a broad range of disorders from a mild soft tissue injury to a severe brain damage leading to death. The head trauma resulted from physical abuse is the main reason for the severe and fatal injuries in children, and arises with the clinical triad of externally seen trauma signs, subdural hemorrhage and retinal hemorrhage. In this article, we report a case of shaken baby aged 20-day, who was referred to our clinic with the diagnosis of late neonatal sepsis and osteogenesis imperfecta. A 20-day-old baby was presented to the emergency department of an outer health center with the complaints of restlessness and non-breastfeeding was considered as sepsis, and subsequently when multiple fractures were detected in the radiological investigations, a diagnosis of osteogenesis imperfecta was also considered, and the baby was referred to our hospital. In the examination of the extremities, he had swelling, ecchymose and limited mobility in both arms and the left calf. The extremity radiograms revealed body fractures in both humeri and the left femur. Radiological investigations are very important for the diagnosis of physical abuse, which causes serious mortality and morbidity and is usually difficult to diagnose because of a wrong medical history given by family members or a history taken by the physician carelessly. Discordance between the history given by the family and the radiological findings should be warning for the physician, and the radiological findings must be assessed carefully.

Key words: Shaken baby syndrome, physical child abuse, newborn, osteogenesis imperfecta, multiple fractures

Yenidoğanda Osteogenesis İmperfekta Tanısı Düşündüren Hırpalanmış Bebek Sendromu

ÖZET

Fiziksel çocuk istismarı, hafif yumuşak doku yaralanmasından ölüme neden olan ağır beyin hasarına kadar değişen geniş bir yelpazede ortaya çıkabilmektedir. Çocuklarda ciddi ve ölümcül yaralanmaların başlıca nedeni fiziksel istismar sonucu oluşan kafa travmasıdır ve başlıca; dıştan gözlenen travma izleri, subdural hemoraji, retinal hemoraji klinik triadı ile ortaya çıkmaktadır. Yazımızda geç neonatal sepsis ve osteogenesis imperfekta tanısı ile merkezimize sevk edilen 20 günlük bir bebekte hırpalanmış bebek sendromu olgusunu sunduk. Yirmi günlük iken huzursuzluk, emmeme şikâyeti ile dış merkez çocuk acil servisine götürülen hasta sepsis düşünülmüş ve bu sırada çekilen graflerinde çok sayıda kırık saptanması üzerine, osteogenesis imperfekta tanısı da düşünülerek hastanemize sevk edilmişti. Ekstremitelerde muayenesinde her iki kol ve sol baldırda şişlik, ekimoz ve hareket kısıtlılığı mevcuttu. Ekstremitelerde graflerinde her iki humerus ve sol femurda cisim kırığı saptandı. Çoğunlukla aile bireyleri tarafından verilen yanlış öykü veya hekimin dikkatsiz aldığı öykü ile tanısı güçleşen, ciddi mortalite ve morbiditeye neden olan fizik istismarın tanısında radyolojik incelemelerin önemi büyüktür. Ailenin verdiği öykü ile radyolojik bulgular arasındaki uyumsuzluk hekim için uyarıcı olmalı ve radyolojik veriler dikkatle incelenmelidir.

Anahtar kelimeler: Hırpalanmış bebek sendromu, fiziksel çocuk istismarı, yenidoğan, osteogenesis imperfekta

¹Department of Pediatrics, Division of Neonatology, Selçuk University, Selçuklu Medical Faculty, ²Department of Pediatrics, Division of Neonatology, Abant İzzet Baysal University, Medical Faculty, Bolu, Turkey, ³Psychiatrist, Department of Psychiatry, Selçuk University, Selçuklu Medical Faculty, Konya, Turkey, ⁴Department of Pediatrics, Division of Neonatology, Konya University, Meram Medical Faculty, Konya, Turkey, ⁵Department of Pediatrics, Division of Neonatology, Konya University, Meram Medical Faculty, Konya, Turkey,

Correspondence: Ali Annagür M.D.
Selçuk Üniversitesi, Selçuklu Tıp Fakültesi, Yenidoğan Servisi, Selçuklu /Konya, Turkey
Tel: +905057906554
E-mail: aliannagur@yahoo.com

INTRODUCTION

Child abuse is a common social and medical issue that leads to serious morbidity and mortality in children. Physical abuse of child is described as intentional infliction of injury, by caregiver, on child leading to bruising, burns, fractures and scratch or organ damage (1,2). According to the United States (USA) National Child Abuse and Neglect Data System (NCANDS), the rate of confirmed physical abuse is 20%. Eighty percent of the offenders are parents. According to the 2002 USA data, the rate of mortality caused by physical abuse is 1.98/100.000 (1400 children). Seventy six percent of the dead children were under 4-year-old, with 41% being under 1-year-old (3). We herein report a 20-day-old newborn that was diagnosed with shaken baby syndrome (SBS) and referred to our clinic with the diagnosis of sepsis and osteogenesis imperfecta.

CASE

A 20-day-old baby was presented to the emergency room of an outer health center with the complaints of restlessness and non-breastfeeding was considered as sepsis, and subsequently when multiple fractures were detected in the radiological investigations, a diagnosis of osteogenesis imperfecta was also considered, and the baby was referred to our hospital. This baby was brought to the hospital by his father. He stated that the baby was admitted to the hospital because of appearing restless, not breast-fed and having spontaneous swellings and bruising in his arms and legs for the last two days. His prenatal, natal and postnatal, and family history indicated no specific finding. His father and mother were graduated from a primary school, and the father had lost his job one week ago. Physical examination in-

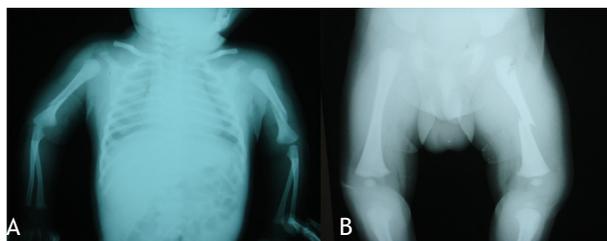


Figure 1. A plain radiogram showing the fractures in both humeri (A) and the left femur (B).

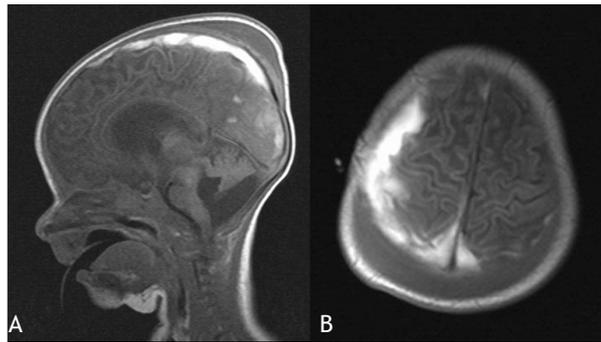


Figure 2. A brain MRI showing the subdural and intraparenchymal haemorrhages (A, B).

dicated a pulse rate of 140 per minute, breath rate of 20 per minute, blood pressure of 83/36 mmHg, weight of 3650g (75-90% percentile), height 52cm (75-90% percentile), head circumference of 36cm (75-90% percentile). His general health status was poor, and he appeared pale, had a bumed frontal fontanelle of 3x3 cm, bilateral palpebral oedema, and bilateral subconjunctival haemorrhage. There was diffuse ecchymoses along the body. Breathing was shallow, and he was bradypneic. In the examination of the extremities, he had swelling, ecchymose and limited mobility in both arms and the left calf and scrotum. He tended to somnolence. Neonatal reflexes were weak. Laboratory findings were as follows: SGOT 41 IU/L, SGPT 34 IU/L, CRP 3 mg/dl (N: <5), white blood cells 8700/mm³, hemoglobin 6.7gr/dl, platelet count 260.000/mm³. Coagulation tests were normal. Lung x-ray imaging was normal, whereas the extremity radiograms revealed body fractures in both humeri and the left femur (Figure 1). During the follow-up period, he was connected to a mechanical ventilator because of the presence of convulsions, worsened consciousness, and deep apneas. Brain magnetic resonance imaging (MRI) revealed subdural and intraparenchymal haemorrhages (Figure 2).

Considering the inconsistency between the physical examination findings and the history and the presence of multiple long-bone fractures along with the brain MRI findings, he was diagnosed with “shaken baby syndrome”. A forensic medicine report was drawn up and presented to the legal authorities. The legal inquiry showed that the case was physically abused by the mother. A psychiatric assessment revealed that the mother had no mental retardation or psychotic condi-

tion, but felt only anxiety and regret due to the event she experienced. The mother was arrested by the court. The case was treated, and then referred to an institution of social services.

DISCUSSION

No large-scale studies have been carried out in order to comprehensively describe the child abuse in Turkey. In general, the studies conducted in Turkey have shown that 14 to 87% of the child population is exposed to physical abuse (1). The risk factors of SBS include premature infants, chronically ill children, babies with colic disorder, children with learning and behavioral problems, stepchild, mental and physical disturbance, single parent, drug addicted parent, having an illegitimate baby, parent having a psychiatric disorder, unemployment and poorness (4-7). History and physical examination (and in some cases laboratory tests and diagnostic imaging techniques) form the basis for diagnosing SBS. Inquiries on the medical history should include the type of birth, perinatal complications, severe bleeding during circumcision, and the history of clinical diseases. A history of osteogenesis imperfecta and coagulopathy should be considered when inquiring the family history. SBS should be considered when the findings obtained from the patient are not consistent with

Table 1. Radiologic Features of Child Abuse.

1. Bucket-handle and metaphyseal corner fracture
2. Multiple rib fractures, especially posterior rib fractures
3. Multiple fractures and/or multiple fractures showing different stages of healing
4. Scapular, acromial, or sternal fractures
5. Transverse, oblique, or spiral fractures of long bones in nonambulatory infants in the absence of significant accidental trauma or fracture inconsistent with given history
6. Fracture separations of distal humeral epiphysis or distal femoral epiphysis (suggestive but nondiagnostic of abuse)
7. Unsuspected fractures found in patient without history of trauma
8. Unexplained fractures associated with normal bone mineralization
9. Lack of repeated fractures in a protective environment
10. Visceral injuries such as duodenal or jejunal hernatomas
11. Head injuries not compatible with mechanism of injury or unexplained, such as subdural hematoma, subarachnoid hemorrhage, diffuse cerebral edema, intracerebral hernatoma, and skull fracture; although head injuries can be the result of child abuse, they are not distinctive and look like abnormalities of other cause

Table 2. Summary of the Salient Features of Osteogenesis Imperfecta

1. Bone fragility, fractures, osteoporosis
2. Blue sclera
3. Deafness or hearing impairment
4. Dentinogenesis imperfecta
5. Wormian bones
6. Positive family history of hearing impairment, dentinogenesis imperfecta, osteogenesis imperfecta, and/or history of fractures or bone deformity
7. Ligamentous laxity and hypermobility of joints
8. Abnormal temperature regulation (heat intolerance, excessive sweating)
9. Easy bruising
10. Fragile skin
11. Short stature, growth retardation
12. Progressive scoliosis
13. Fractures continue in protected environment

the history given by the family (7-12). For the physical abuse, the most frequently observed sign on skin is bruising. Bruising caused by an accident is usually seen on the front surfaces of legs, and in the knees and forehead. Bruising on the genital organs, dorsal areas, and the dorsal surface of hands should be considered doubtful. In addition to bruising, burns, lacerations, bites, and cuts may be seen. In case of intentional fluid burns, lesions due to immersion, submersion or splashing occur. Submersion burns have sharp borders; the glove and sock type lesions occur in the extremities. In the accidental burns, there are many splashing burns with irregular borders (13). In SBS, the most frequent cause for death is head trauma. During the first year of life, 95%

Table 3. Features in Clinical History and Physical Examination Helpful to the Radiologist in Diagnosing Child Abuse

1. Injury with cause or extent not compatible with a given history
2. Inconsistent or conflicting history
3. Lack of history of significant trauma in patient with fractures
4. Lack of or delay in seeking appropriate medical attention for an injury
5. Retinal hemorrhages
6. Specific bruises (e.g., palmprints, fingerprints, strap marks), bruises in unusual places (perineum), human bites, and cigarette burns
7. Lack of blue sclera
8. Lack of clinical features or family history of osteogenesis imperfecta (see Table-2)

of the head traumas are intentional injuries. One fourth to half of the children presented to hospitals due to a head trauma caused by maltreatment are children under the age of 24 months (14). A head trauma should be suspected if a baby had coma, convulsion, apnea, bulging fontanelle or increased intracranial pressure (8,15). Subdural hematoma accompanies the clinical picture in 38 to 100% of the cases and usually occurs bilaterally (2,4,9,16). Spinal cord injury is generally accompanied by the long-bone and costal fractures. These cases may be presented with symptoms such as lethargy, vomiting, restlessness, loss of feeding, non-breast-feeding, and difficulty in breathing (17). Of the SBS cases with subdural hematoma, 75 to 90% are accompanied by uni- or bilateral retinal haemorrhage (9,15-18). Brain MRI is superior to brain CT in showing the lesions in detail and diagnosing the head trauma (16,17). Our case had subdural and intraparenchymal haemorrhages in brain MRI. In SBS, the second most frequent cause for death is intraabdominal injury due to hits. The affected children may experience repeated vomiting, abdominal swelling, loss of intestinal sounds, localized tenderness, and shock (3,8,13).

A skeletal damage is often seen in trauma not due to accidents. Some fractures, metaphyseal lesions (bucket stem or corner fractures), posterior costal fractures, scapular fractures, spinous process fractures and sternal fractures may possibly indicate child abuse. Multiple fractures (at different stages of healing), epiphyseal detachments, vertebral body fractures, complex cranial fractures and finger fractures are moderately suspected. In a baby who can not walk, spiral femur fractures and non-supracondylar humerus fractures should especially suggest non-accidental traumas (19). Radiological features of the physical child abuse are summarized in Table-1 (20). In addition to the complexity of the diagnosis of an abuse, similar skin lesions, bruising or fractures may occur in many cases. The most striking case is osteogenesis imperfecta. The diagnosis of abuse can be confused with this type-1 collagen disturbance. Moreover, a case of osteogenesis imperfecta can be considered abuse by mistake, leading to tragic consequences (21). In case of osteogenesis imperfecta, osteopenia, fine cortex, and deformities in long bones caused by the improving fractures can be detected radiologically (22). The wormian bones in cranium, blue sclera, loss of hearing, dentogenesis imperfecta, and a family history of osteogenesis imperfecta may be helpful in diagnosing

(22,23). The main features of osteogenesis imperfecta are summarized in Table-2 (20). It is important to remember that a case of underlying osteogenesis imperfecta would not rule out the possibility of abuse. The main clinical history and the characteristics of a physical examination are given in Table-3 to help the physician in the diagnosis of physical abuse (20). A physician should not avoid reporting the suspected cases of abuse while waiting the results of laboratory tests (9,24,25).

The differential diagnosis should also take into consideration hemophilia, trauma caused by a real accident, Caffey disease, osteomyelitis, premature osteopenia, congenital insensitivity to pain, copper deficiency, leukemia, rachitis, congenital syphilis, and birth trauma. Some renal diseases and drug reactions may also cause changes in bone and suggest child abuse. A detailed anamnesis, physical examination, laboratory testing and radiological investigation are all helpful for the diagnosis and differential diagnosis (1,13,20,23,24). In the plain radiograms, there were no bone abnormalities such as osteopenia pertaining to the fractures caused by osteogenesis imperfecta, blue sclera and the relevant family history (22). Our case had been referred to a tertiary health centre with the diagnosis of sepsis and osteogenesis imperfecta as a result of inappropriate history, physical examination and laboratory investigations in the centre where the case was seen first. However, SBS was suspected because of the discordance between the current findings and the history. The diagnosis of SBS was confirmed by the presence of diffuse ecchymoses along the whole body, ruling out the infectious and hematological diseases with the laboratory findings, the presence of multiple long-bone fractures in the plain radiograms, a poor neurological status, and the detection of the intracranial and subdural haemorrhages in the brain MRI, which was obtained because of the occurrence of a retinal haemorrhage. As a conclusion, it should be kept in mind that the cases of SBS would be confounded by the systemic diseases including sepsis, osteogenesis imperfecta, and might be seen with various clinical pictures. The possibility of child abuse may be overlooked in pediatric emergencies. A radiological evaluation is very important for diagnosing SBS. Therefore, it is essential for an early diagnosis and treatment that the physicians working in pediatric emergency rooms take a careful anamnesis and perform a complete physical examination and radiological evaluation, considering SBS in the differential diagnosis.

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