Genetic and Congenital Defect Conditions That Mimic Child Abuse

Terrance D. Wardinsky, MD
Sacramento, California

Several medical conditions have been reported to mimic the physical manifestations of child abuse and neglect (CAN). These conditions include genetic, congenital, and other disorders that may result in poor weight gain, bone fracture, or skin lesions that appear to be bruises or burns. Society demands that medical personnel who care for children be aware of the many indicators that suggest CAN. This article is a review of 6 of 18 cases among 264 that were referred to a child abuse team over a 3-year period because of suspected abuse in which medical disorders were initially and erroneously diagnosed as CAN. This report also suggests ways to support aggrieved parents who have been mistakenly reported to be child abusers.

Key words. Child abuse; diagnostic errors; child welfare; genetic; abnormalities. (*J Fam Pract 1995; 41:377-383*)

Child abuse must always be suspected in a child with unusual marks or injuries that do not match the given history.[1] Many pediatric conditions that resemble child abuse and neglect (CAN) have been reported in the literature.[2,3] Over several months, as a congenital defect consultant at a medical center, I have consulted on a number of cases referred to a child abuse team that, on further assessment, were found to involve genetic or congenital defect conditions rather than child abuse. This paper briefly discusses these cases and reviews the current literature addressing genetic and congenital conditions that have been mistakenly diagnosed as CAN. Once a mistaken diagnosis has been corrected and CAN ruled out, supportive parental guidance should be provided.

Case Reports

Patient 1

A 10-month-old infant presented after having "climbed out of her crib," falling and striking her head on a hardwood floor. Assessment revealed a contused left parietal region with a notable linear skull fracture of the left parietal bone crossing to the frontal region (Figure 1, *left*). A follow-up skeletal survey revealed three calloused and healing rib fractures that were older than the skull fracture (Figure 1, *right*). Child abuse was suspected. Although the parents were consistent in explaining the circumstances of the alleged accident and seeking medical care, the child was placed in foster care for protection. The skeletal survey was reviewed by a pediatric radiologist, who noted osteoporosis of the bony architecture. On further genetic consultation it was revealed that the child had unusual slate-blue sclera; she was short and had a foot deformity. A skin fibroblast biopsy was performed because of the possiblity of osteogenesis imperfecta (OI); subsequently, type IV osteogenesis imperfecta was proved by molecular DNA collagen analysis. The child was returned to her parents after 2½ months in a foster home. Neither the parents nor the sibling showed evidence of collagen abnormality by skin fibroblast study. One of the unfortunate consequences of this misdiagnosis was the accumulation of court costs resulting from the distraught parents' attempts to have their child legally returned.

Features that may help differentiate OI from child abuse include: frequent fractures, radiographic evidence of osteoporosis, short stature with a frontal boss of the head, bowed extremities, discolored sclera and teeth, and

Submitted, revised, May 25, 1995.

This research was presented as a poster at the March of Dimes Conference on Clinical Genetics held at Stanford University, Stanford, California, July 12–13, 1992.

The study was conducted while Dr Wardinsky was the Congenital Defect Consultant to the Departments of Pediatrics and Family Practice, David Grant USAF Medical Center, Travis Air Force Base. Requests for reprints should be addressed to Terrance D. Wardinsky, MD, Alta California Regional Center, 2031 Howe Ave, Suite 100, Sacramento, CA 95825.

ISSN 0094-3509

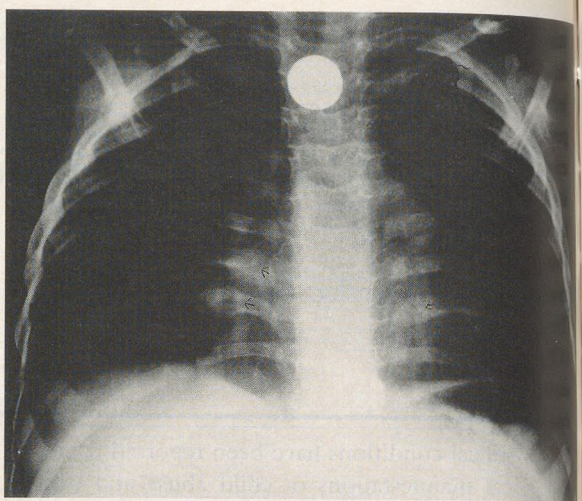

Figure 1. *Left.* Note the skull fracture on this radiograph of a patient with osteogenesis imperfecta type IV as described in text as patient 1. *Right.* Note posterior rib fractures on this radiograph of patient 1.

occasionally a positive family history for this condition. Osteogenesis imperfecta type IV can be either a spontaneous mutation or inherited as an autosomal dominant disorder.

Patient 2

A 3-year-old girl with a contusion and swelling on the right side of her face was referred by the child care center for suspected CAN. The child care worker was concerned because it appeared that the child had been struck in the face. Physical examination revealed a healthy appearing child who was in no acute distress. The child appeared to have a swollen right cheek and neck with a generalized discolored blue hue and some blue-red marks within the central area (Figure 2). Closer examination suggested that these apparent injuries were instead a facial vascular malformation that had progressively infiltrated the child's face since early age. This suspicion was confirmed by the mother and medical record documentation. No further assessment was performed; however, the parent was significantly upset by the implication of child abuse.

Vascular marks such as hemangiomas usually are present from birth (congenital) or become evident shortly after birth. They usually are circumscribed with defined borders and blanch with pressure. Typical cavernous hemangiomas involute in time as they outgrow their vascular supply. In contrast to cavernous hemangiomas, vascular malformations, as in this patient, tend to persist and may even enlarge. Both are sporadic events and generally are not inherited.

Patient 3

On a well-baby examination, a 7-month-old female infant was noted to have a head circumference that had increased beyond 2 standard deviations and a bulging fon-

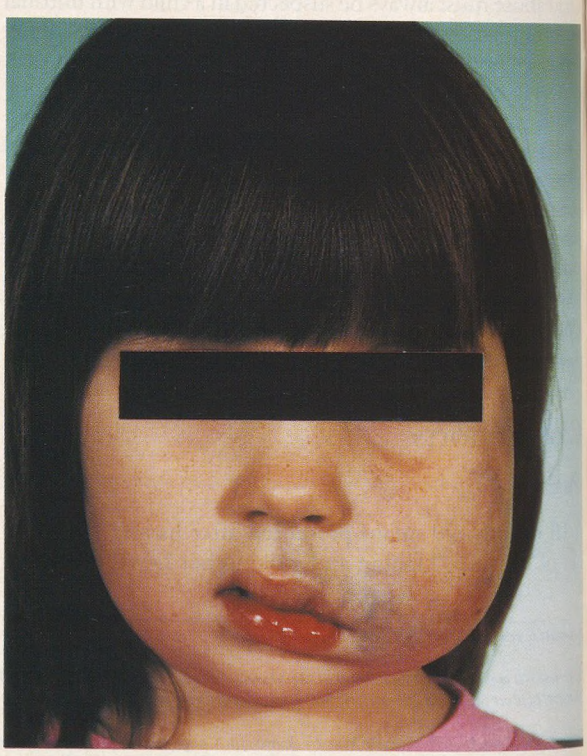

Figure 2. Vascular malformation of face with facial suffusion that was misinterpreted by day-care personnel as possible blunt trauma to face.

tanelle with a occipital-parietal boss. The child was referred for further workup with the history of occasional lethargy. A head computerized tomographic scan was performed and was initially interpreted as showing bilateral subdural hematomas of undetermined cause. Further assessment for possible infant whiplash from "shaken baby" syndrome was undertaken; results of the skeletal series, retinal examination, and coagulation studies were unremarkable. History failed to reveal previous trauma. The family was referred to the Child Protective Service (CPS) for home environmental and social evaluation. Indecision and delay in CPS home evaluation in this case resulted in prolonged admission for protective observation. The parents were very angry over the initial incrimination. A pediatric radiology consultation suggested further central nervous system (CNS) imaging, including a brain magnetic resonance imaging study and ultrasound of the brain surface. These studies helped rule out traumatic hemorrhage by demonstrating subarachnoid space enlargement with chronic cerebrospinal fluid (CSF) collection compatible with benign external hydrocephaly of infancy, also known as benign communicating hydrocephalus of infancy (Figure 3). This condition is generally self-limiting and frequently regresses spontanously.

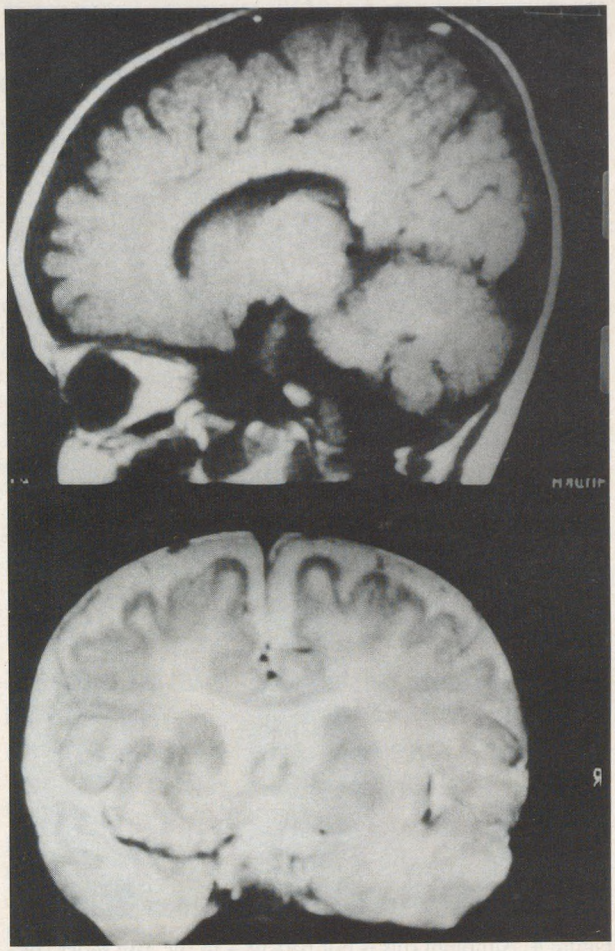

Figure 3. Benign hydrocephaly of infancy (communicating hydrocephaly) with rim of cerebrospinal fluid (CSF) extending around the cerebral cortex was initially misinterpreted as subdural hematoma. Brain ultrasound and magnetic resonance imaging may distinguish meningeal spaces as well as density of fluid to separate CSF from hemorrhage.

Patient 4

A 4-year-old girl with ecchymoses and scars predominantly over the lower extremities but with some on the upper right hand and forearm was referred by a babysitter. The child was evaluated in the pediatric clinic, and because of no explanation other than she "seemed to bruise easily," the child was admitted to the pediatric ward for CAN observation. Family history was did not reveal any bleeding or skin disorders. Coagulation studies and radiographs were normal. The CPS agency evaluated the home but could not find significant dysfunction or CAN. Further consultation revealed that the child had very smooth, lax skin. The child also had excessive joint mobility. Although not biochemically proven, she was thought to have Ehlers-Danlos syndrome types I or II. Because the family subsequently left the military, no follow-up was available.

Connective tissue disorders classified as Ehlers-Danlos types I to X involve laxity of skin and joints. Some of the subtypes have a propensity for poor healing with thin, cigarette-paper–like scar (papyraceous) formation as well as bruising (Figure 4), and prolonged bleeding and occasional vascular disruption. Tissues of some patients hold sutures poorly. Ehlers-Danlos syndrome types I and II are inherited in an autosomal dominant fashion.

Patient 5

The babysitter of a 4-month-old Hispanic male infant called the CPS agency because of concerns about physical abuse. CPS arranged for further medical assessment of the infant, which revealed blue mongolian pigmentary marks over the buttocks, back, and upper thighs (Figure 5). Although the episode was embarrassing to the parents, who accompanied the child to the clinic with the CPS social worker, they were understanding after further explanation regarding required reporting procedures as well as the normal occurrence of these birthmarks. Mongolian spots or congenital blue marks are common to infants of pigmented races. These marks often can be found on the back, buttocks, and posterior extremities. They usually

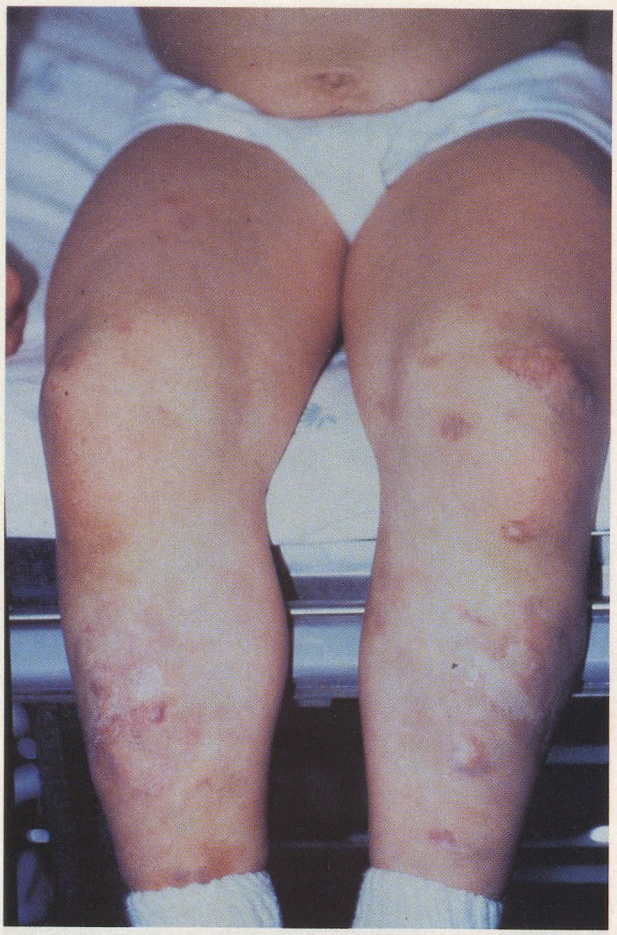

Figure 4. Although this is not patient 4 in the case series (pictures not available), this patient has Ehlers-Danlos syndrome type I with typical bruises and scars and unusual healing of skin (papyraceous) that could easily be interpreted as marks of child abuse and neglect.

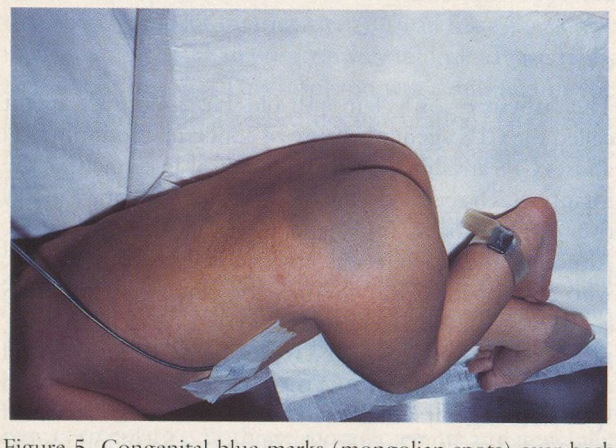

Figure 5. Congenital blue marks (mongolian spots) over back and buttocks that could be mistaken for bruises.

are of a consistent color, have defined borders, and tend to disappear with age.

Patient 6

The lifeguard at the community swimming pool referred to the CPS a young child who appeared to have multiple bruises on her extremities. The child was evaluated in our clinic, and on further inquiry and record check, a previous diagnosis of Shwachman-Diamond syndrome was revealed. This autosomal recessive disorder is associated with short stature, developmental delay, exocrine pancreatic insufficiency, short ribs with metaphyseal dysplasia, and isolated or combined decreases in red cell, white cell, or platelet counts. The patient recently had lowered platelet counts with increased bruising. Further social and family assessment did not reveal concerns for CAN. The family was embarrassed by this mistaken diagnosis but understood the nature of the referral. Conditions that affect the marrow elements as well as the coagulation cascade may present with bruising and recurrent episodes of bleeding. A history of excessive bruising is an indication to rule out bleeding disorders.

Discussion

Making the Correct Diagnosis

Society demands that medical personnel who care for children be aware of the many indictors that suggest CAN. To prevent a mistaken diagnosis, however, these caregivers must also be able to recognize that many pediatric medical and physical conditions can mimic CAN. With society's increased vigilance, overreporting of conditions that resemble CAN is possible, expecially in the absence of a complete assessment. This problem is actually encouraged by civil law, which requires only that abuse is a probable diagnosis.[4]

To determine the frequency and diversity of misdiagnosed CAN resulting in false accusations of parents or child-care providers, a 36-month retrospective review of child advocacy case records from January 1, 1988, to December 31, 1991, was performed at David Grant Medical Center, Travis Air Force Base, in California. Cases referred to our CAN team in accordance with US Air Force Regulation 130–68 are classified as substantiated or unsubstantiated. Cases are substantiated when an offender confesses and when CPS or a court determines that maltreatment occurred. Other consideration toward substantiation is given to the reliability of a witnessed event, the reliability of the victim reporting CAN, and the opinion of an expert in CAN that maltreatment occurred.

Table. Genetic and Congenital Defect Conditions Reported in the Literature as Mimicking Child Abuse and Neglect

Genetic or Congenital Condition	Signs and Symptoms that Mimic Child Abuse and Neglect
Muscular dystrophy[2]	Leg fractures
Epidermolysis bullosa[6]	Blisters and scars
Osteogenesis imperfecta[7]*	Osteoporosis, fractures, blue sclera
Ehlers-Danlos syndrome[8,9]*	Bruises, atropic skin scars, blue sclera, skin and joint laxity
Menke kinky hair syndrome[10]	FTT, fractures
Mucopolysaccharidoses/lipidoses[11,12]	FTT
Congenital insensitivity to pain disorders[12]	Skin trauma
Rickets[12–14]	Osteoporosis, fractures, bone pain
Meningomyelocele[12,14]	Leg fractures
Immune incompetence disorders[15]	FTT, recurrent infection
Cystic fibrosis[16]	FTT, bleeding
Myotonic dystrophy[17]	Anal laxity similar to sexual abuse
Bony dysplasias[18,19]	Fractures, short stature
Hemophilia[2,20]	Bruises, skin trauma
von Willebrand's disease[21]	Bruises, skin trauma
Mongolian spots[22]*	Bruises, skin trauma
Hemangioma and vascular malformations[23]*	Bruises, skin trauma
Schwachman-Diamond syndrome*	Bruises
Crohn's disease	Chronic abdominal pain
Benign external hydrocephaly*	Rapid head growth and bulging fontanelle

**Cases described in this report.*
FTT denotes failure to thrive.

Without meeting these criteria, the case is considered unsubstantiated. In the 36-month period during which 504 cases were referred, 264 were unsubstantiated. Eighteen of the 264 unsubstantiated cases were determined, after subsequent complete assessment, to have initially received an erroneous diagnosis of CAN. Eight of these 18 (44%) had established diagnoses of genetic or congenital defect conditions, as substantiated by a review of medical records. Six of the 8 patients were subjected to extensive CPS evaluations. The remaining 10 of the 18 patients were found to have common pediatric diagnoses, eg, impetigo, fungal skin infections, sunburn, stocking constriction marks, that initially were thought to be markers of CAN. These 10 patients had either minimal or no CPS referral.

Although in the past decade there have been several reports of misdiagnosed cases of CAN and the conditions that mimic abuse, no specific population data are currently available to estimate the true number of CAN false reports. Based on this retrospective review, it appears that the incidence of misdiagnosis is rather high: 18 (7%) of 264 unsubstantiated cases, or 18 (3.5%) of 504 if both substantiated and unsubstantiated cases are included. One reason for the higher rate in this study of a military population may be overreferral secondary to the ready access to and availability of physicians and social workers afforded by the military's mandated Family Advocacy Programs. However, it does not appear that the rate of CAN is any higher in the military than in civilian populations.[5]

Many inherited connective tissue disorders can lead to a mistaken diagnosis of CAN because patients present with poor weight gain and failure to thrive or with skin lesions that appear as bruises, burns, and bone injury. Approximately one half (8/18) of the cases in this report were found to involve genetic or congenital defect conditions that involved skin or skeletal findings rather than physical abuse. The Table lists many of the signs and symptoms of genetic and congenital conditions that, because of their nature and subtle presentations, have been mistaken as abuse. Several conditions, such as skin, skeletal, and connective tissue disorders, hematological and coagulation diseases, musculoskeletal ailments, metabolic disorders, and congenital defects, have been previously reported. Because cases involving these systems can be confusing, further consultation with a child advocacy specialist or geneticist may be helpful to rule out these conditions. Bone density, specific fracture types indicative of CAN (ie, rib, epiphyseal-metaphyseal, vertebral compression, periostitis, and lateral clavicle), patterns of distribution, and different stages of healing are hallmarks that help separate inheritable bone and connective tissue disorders from traumatic bone and skin lesions typically associated with child abuse.

Benign external hydrocephaly, also known as communicating hydrocephaly or Schwachman-Diamond syndrome, has not previously been reported in the literature as mimicking CAN. Benign external hydrocephaly, which can be confused with CNS injury, may be differentiated by CNS imaging studies, such as computed tomography, magnetic resonance imaging, and ultrasound, to separate densities of the meningeal layers and spinal fluid from traumatic injuries that result in the collection of blood.[24]

The worst of all scenarios occurs for parents of infants and children who die suddenly and unexpectedly of an inheritable disorder and whose injuries or illness may have initially been misdiagnosed as CAN. A recent example of this was presented on a television movie, *Without a Kiss Goodbye,* based on a true story of metabolic disorder methylmalonic aciduria presenting as severe failure to thrive and death in a young infant. The mother, after a trial, was imprisoned for CAN. While in prison, she gave birth to another child who, immediately placed in foster care, deteriorated in a fashion similar to that of the sibling. On more extensive workup, the second infant proved to have methylmalonic aciduria and was treated. The mother and family were eventually exonerated.

In some circumstances, the failure to properly diagnose may allow a treatable disorder, eg, hemophilia or osteogenesis imperfecta, to go unrecognized. On the other hand, it is worth remembering that children with these unusual conditions could also be victims of CAN.[6]

Supporting Wrongly Accused Parents and Caretakers

Physicians who inadequately assess or inappropriately refer suspected cases may produce emotional harm to the misdiagnosed child and family and potentially may be liable for incomplete evaluation prior to reporting.[25] Inappropriate reporting may do irreparable harm to the patient-physician relationship. Civil and criminal charges and subsequent foster home placement may follow. Several cases reported in this review involved extensive CPS evaluations with prolonged hospital stays for protective observation. Unfortunately, a child may be placed within the foster system, as occurred with the first patient in our series. This child was removed from her family for 2½ months prior to the skin fibroblast biopsy that proved the diagnosis of osteogenesis imperfecta type IV. In addition to losing the care of a mother or father or both, a child in the foster care system may be shifted from home to home. An aggrieved parent may lose trust in health professionals to the extent of hiding genuine accidental trauma for fear of being reported.[26]

Once a mistaken diagnosis has been made, it may be difficult to repair the parental anguish of having been "reported as a child abuser." We have identified several methods that can be helpful in supporting parents who are falsely accused: (1) an apology to the parent(s) by those making the report, (2) an explanation of the child protective process that is geared to overreporting in an effort to protect children, (3) individual and family counseling, including education about the newly discovered diagnosis, (4) formal psychologic counseling, (5) participation in educational conferences and talking with other families mistakenly reported as perpetrators of CAN. It also is especially important to counsel children that the unfortunate series of events related to the false report of CAN is not "their fault."

Conclusions

Several genetic and congenital defect conditions have been presented that may be misdiagnosed as CAN. Physicians who evaluate children for possible CAN must be aware of the many genetic and congenital defect signs and symptoms, eg, bone, hematologic, skin, and metabolic conditions, that can mimic CAN. As clinicians, we must completely assess all the circumstances and appreciate the complete differential diagnosis of these disorders that may be mistaken for CAN in order to prevent the anguish associated with mislabeling caretakers as child abusers. If, after appropriate evaluation including consultation, it is still unclear whether child abuse or neglect is involved, it is better to report suspected CAN than to do nothing, as the consequences of failing to detect and report CAN are far worse than those of mistakenly diagnosing CAN.

References

1. Kempe CH, Silverman FN, Steele BF, Droegemueller W, Silver HK. The battered-child syndrome. JAMA 1962; 181:17–24.
2. Wheeler DM, Hobbs CJ. Mistakes in diagnosing non-accidental injury. 10 years' experience. BMJ 1988; 296:1233–6.
3. Kirschner RH, Stein RJ. The mistaken diagnosis of child abuse. A form of medical abuse? Am J Dis Child 1985; 139:873–5.
4. Krugman RD. Advances and retreats in the protection of children. N Engl J Med 1989; 320:531–2.
5. Dubanoski RA, McIntosh SR. Child and neglect in military and civilian families. Child Abuse Neglect 1984; 8:55–67.
6. Colver GB, Harris DW, Tidman MJ. Skin diseases that may mimic child abuse. Br J Dermatol 1990; 123:129.
7. Carty H. Brittle or battered. Arch Dis Child 1988; 63:350–2.
8. Pope FM, Narcisi P, Nicholls AC, Liberman M, Oorthuys JW. Clinical presentations of Ehlers-Danlos syndrome type IV. Arch Dis Child 1988; 63:1016–25.
9. Roberts DI, Pope FM, Nicholls AC, Narcisi P. Ehlers-Danlos syndrome type IV mimicking non-accidental injury in a child. Br J Dermatol 1984; 111:341–5.
10. Adams PC, Strand RD, Bresnan MJ, Lucky AW. Kinky hair syndrome: serial study of radiologic findings with emphasis on the similarity to the battered child syndrome. Radiology 1974; 112: 401–7.
11. Adebahr G, Ritter C. Mucopolysaccharidose II (Hunter-Syndrom) und III (Sanfilippo-Syndrom) als Ursache einer zunachst auf Vernchlassigung bezogenen Abmagerung. Z Rechtsmed 1985; 95: 285–95.
12. Merten DF, Carpenter BL. Radiologic imaging of inflicted injury in the child abuse syndrome. Pediatr Clin North Am 1990; 37:815–37.
13. Gefter WB, Epstein DM, Anday EK, Dalinka MK. Rickets presenting as multiple fractures in premature infants on hyperalimentation. Radiology 1982; 142:371–4.
14. Kleinman PK. Diagnostic imaging of child abuse. Baltimore, Md: Williams & Wilkins, 1987.

15. Copeland AR. A case of panhypogammaglobulinemia masquerading as child abuse. J Forensic Sci 1988; 33:1493–6.
16. Carpentieri U, Gustavson LP, Haggard ME. Misdiagnosis of neglect in a child with bleeding disorder and cystic fibrosis. South Med J 1978; 71:854–5.
17. Reardon W, Hughes HE, Green SH, Lloyd Woolley V, Harper PS. Anal abnormalities in childhood myotonic dystrophy—a possible source of confusion in child sexual abuse. Arch Dis Child 1992; 67:527–8.
18. Herbich VJ, Holczabek W, Lachmann D, Zweymuller E. Zur Differentialdiagnose der Kindesmibbandlung. Beitr Gerichtl Med 1973; 31:97–101.
19. Kleinman PK. Schmid-like metaphyseal chondrodysplasia simulating child abuse. Am J Roentgenol 1991; 156:576–8.
20. Schiver W, Brueschke EE, Dent T. Hemophilia. J Fam Pract 1982; 14:661–2, 664.
21. O'Hare AE, Eden OB. Bleeding disorders and non-accidental injury. Arch Dis Child 1984; 59:860–4.
22. Asnes RS. Buttock bruises = mongolian spots [letter]. Pediatrics 1984; 74:321.
23. Williams CM, Spector R, Brown M. Cervical bruises—a battered child? Cystic lymphangioma. Arch Dermatol 1986; 122:1069–70.
24. Kendall B, Holland I. Benign communicating hydrocephalus in children. Neuroradiology 1981; 21:93–6.
25. Kaplan JM. Pseudoabuse—the misdiagnosis of child abuse. J Forensic Sci 1986; 21:93–6.
26. Qureshi B. Multicultural aspects of child abuse in Britain. J R Soc Med 1989; 82:65–6.