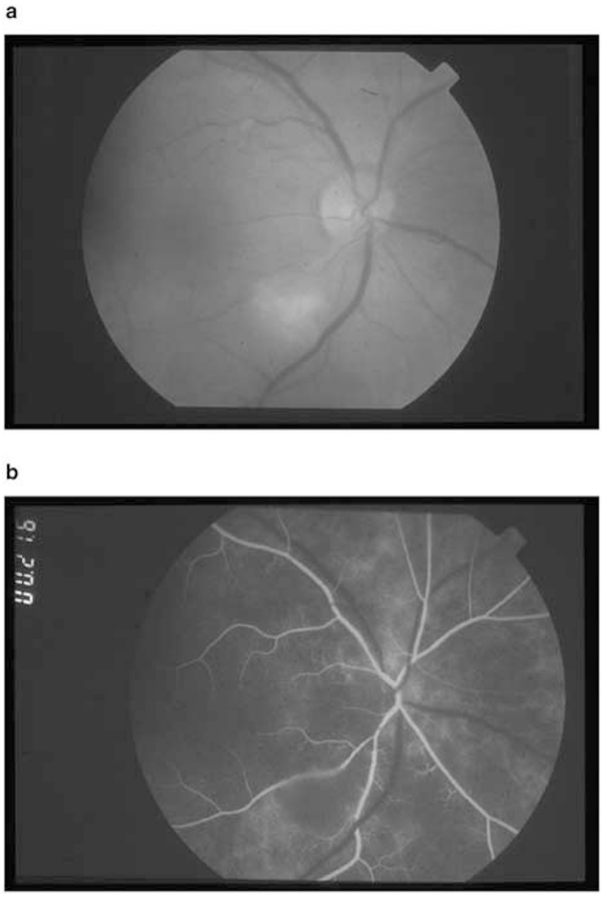

Figure 1 (a) Cotton wool spots of the right fundus. (b) Right fundus fluorescein angiogram at 22 s. The background choroidal fluorescence is still very patchy, indicating marked hypoperfusion.

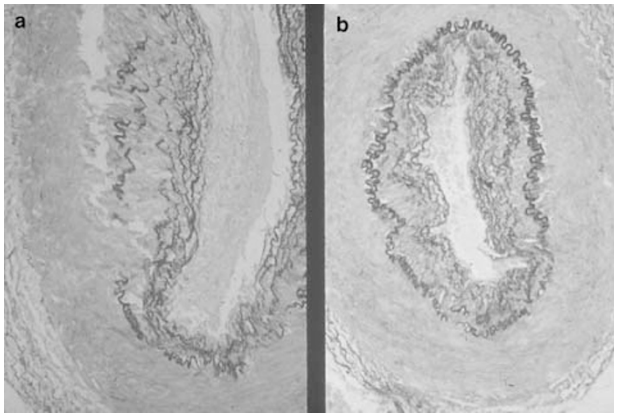

Figure 2 (a) Patient's temporal artery showing disruption of the elastic lamina and of the muscle by giant cell arteritis. (b) Control patient temporal artery biopsy showing arteriosclerosis only. Stain—Elastic van Gieson. Magnification $\times 100$.

both the retina and choroid found on angiography added weight to a diagnosis of vasculitis, particularly giant cell arteritis,[6] and led to a temporal artery biopsy. Clinical suspicion was confirmed with a positive biopsy, despite both normal ESR and CRP.

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Sir,

The role of an ophthalmologist in suspected non-accidental injury

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The diagnosis of paediatric nonaccidental injury (NAI) can have major implications for the future of a child and

the parents.[1] While it is associated with a multitude of systemic and ophthalmological signs, several conditions may masquerade as NAI, including connective tissue and haematological disorders.[2–4] However, an ophthalmologist's role is crucial to avoid misdiagnosis and inappropriate management. We thus report a 2-month-old baby with an initial clinical picture highly suggestive of NAI, but with an underlying diagnosis of osteogenesis imperfecta (OI) that was reached only after a comprehensive ophthalmological evaluation.

Case report

A 2-month-old baby girl was admitted by the paediatricians with a 2-day history of bilateral thigh swelling and distress. Her previous medical, family, and social history was unremarkable with no record of birth trauma, bone or connective tissue disorder. Importantly, there was no history of trauma witnessed by her parents. Clinical examination revealed a right swollen tender upper thigh with increased tone and a reduced range of movement in all directions at the hip joint. An X-ray of her upper legs revealed a spiral fracture of the right femoral shaft (Figure 1). A skeletal survey including a CT scan was carried out and opinion was sought from a group of consultant radiologists. The findings were considered to be highly suggestive of NAI, although the rest of her systemic examination was unremarkable. A multidisciplinary meeting involving the medical and nursing teams, social services, and police was promptly held. In the best interests of the child, parental access was officially restricted and allowed only under supervision.

An ophthalmological opinion was sought to confirm the diagnosis of NAI 2 days following admission. This was initially performed in a darkened room with a dilated pupil. Indirect ophthalmoscopy was unremarkable, with no visible retinal haemorrhages, providing no direct ocular evidence of NAI. In view of the presumed diagnosis of NAI, an opinion was sought from a consultant ophthalmologist and the baby was reviewed in daylight the next morning.[4] The retinal examination was again normal, but on this occasion the child was clearly noted to have blue sclerae bilaterally (Figure 2). The finding of femoral fractures (Figure 3) with blue sclerae was highly suggestive of OI, this being confirmed by a paediatric radiologist on review of the skeletal survey X-rays.[5] These showed characteristic features of generalized osteopenia, abnormal thinning of cortical bone, and the presence of multiple small bone islands within cranial suture lines (Wormian bones).[6,7] The parents were immediately informed of the revised diagnosis of OI and given full access to the baby.

Comment

NAIs can present with a spectrum of systemic and ophthalmological manifestations.[2] A history of multiple injuries on separate occasions, with a characteristic delay in seeking medical care, is often observed. Systemic findings can include surface ecchymoses, multiple fractures, unusual scalds, burns, and injuries that are inconsistent with the history.

Ocular findings in NAI may vary according to the force used and the type of injury sustained by the victim.[2,4] While the most common sign is the presence of peripheral retinal haemorrhages, other findings can include periocular bruising, subconjunctival haemorrhages, hyphaema, dislocated or partially subluxated lenses, retinal haemorrhages, and retinal detachment. Importantly, none of these signs were present in our patient, making the diagnosis of NAI unlikely.

Uniformly blue sclerae are classically found in OI, but may also be seen in Ehlers–Danlos syndrome and keratoglobus, while a focal blue scleral appearance can be caused by a staphyloma.[8,9] Although unexplained

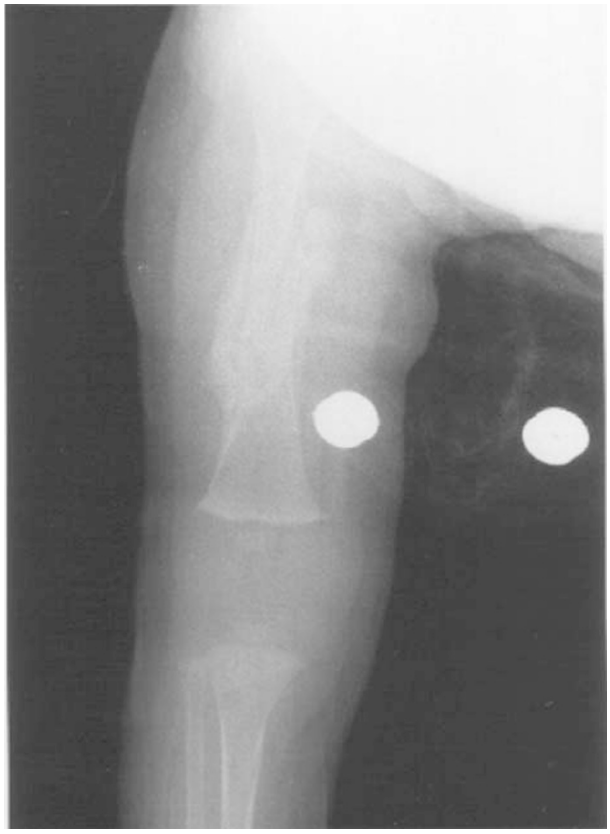

Figure 1 X-ray showing spiral fracture of the femur with slight periosteal reaction.

Figure 2 Patient's appearance with obvious blue scleral.

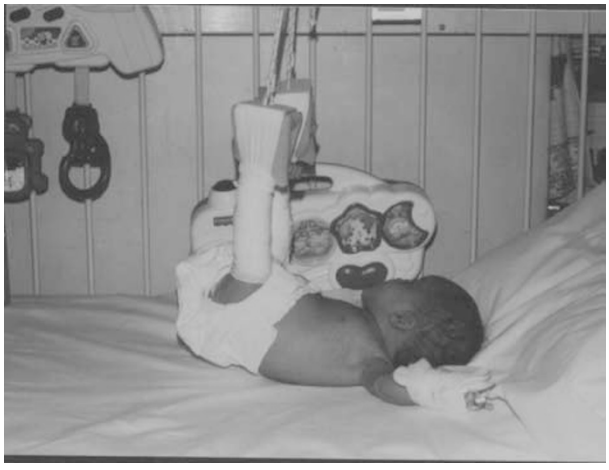

Figure 3 Photograph of baby with right femoral fracture and bilateral sling fraction.

fractures may also be caused by NAI, the finding of blue sclerae with multiple fractures is strongly suggestive of OI, as was found in our patient.[10]

OI is an autosomal dominant inherited disorder characterized by increased bone fragility and progressive bone deformity, with up to six subtypes having been described.[5] Mutations in the genes encoding for pro-alpha 1(1) and pro-alpha 2(1) collagen are present, leading to defective synthesis of type I collagen chains.[11] This leads to deformities of bones, which are brittle and fracture easily with minimal trauma, abnormalities of skin, teeth, tendons, deafness and blue sclerae. Scleral collagen fibres in OI type III are 50% narrower than normal age-matched controls, and are much more uniform in size.[12] This is thought to lead to scleral translucence, causing it to appear blue due to the underlying choroid. Although genetic testing is available for OI, the diagnosis is essentially clinical.[5]

Our case clearly demonstrates the vital role of the ophthalmologist in reaching the correct diagnosis in cases of suspected NAI. This is particularly important, since the misdiagnosis of paediatric NAI is ultimately detrimental to the well-being of both the child and its family. In particular, the ophthalmic examination must always be complete in these instances and not merely restricted to fundoscopy, but must include the whole globe and ocular adnexae.

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