Brief Report

Right Mandible Swelling of Unknown Origin

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Infantile cortical hyperostosis (ICH), or Caffey's disease, has a low prevalence, is not easily recognized clinically, and is seldom reported in the primary care literature. A case of infantile cortical hyperostosis of the right mandible of a newborn is reported. Multiple radiographic tests, as well as bone biopsy, were necessary to establish a definitive diagnosis. The most striking features of this disease are presented, including the clinical and physical presentation, radiographic characteristics, and pathology.

Key words. Hyperostosis, cortical, congenital; facial bone; mandible. (*J Fam Pract 1996; 42:401-403*)

Infantile cortical hyperostosis (ICH), or Caffey's disease, is a rare entity of unknown origin that primarily affects infants. There are no pathognomonic findings, but certain characteristics are frequently present, such as facial bone involvement, particularly involvement of the mandible, which occurs in 80% of cases. Although the degree and extent of bone involvement may vary, there is always a component of inflammation.[1–5] Several cases have been reported among members of the same family, suggesting genetic transmission of the disease, but other theories advocate an immunologic or a viral origin.[6,7]

The diagnosis of this disease is obscured by the general nature of the signs and symptoms. The patient may present with signs and symptoms such as fever, subtle bony swelling, pain, erythema, poor appetite, failure to thrive, and, occasionally, dysphagia. The diagnosis should be established based on the clinical history, radiographic findings, and, if indicated, anatomic pathology.[1–5,8–10] The differential diagnosis includes infection (osteomyelitis), neoplasm, and benign tumors. Because of the low incidence and prevalence of this disease in the 1990s, few physicians are acquainted with it and the diagnosis is usually one of exclusion.

Case Report

A 24-day-old infant presented to the hospital with a temperature of 103°F, a history of fussiness, and extensive oral thrush. The child had an unremarkable past medical history. The patient's mother was healthy, received adequate prenatal care, and had an uneventful pregnancy and delivery. Because of the child's constitutional symptoms and fever, a septic workup was done and the patient was admitted for therapy with intravenous ampicillin and gentamicin. The workup included chest radiography, complete blood count and differential, erythrocyte sedimentation rate (ESR), lumbar puncture, and cultures of the cerebrospinal fluid, urine, and blood.

On the second hospital day, the child was noted to have bilateral parotid gland swelling, but this was thought to be viral, secondary to the child's illness. By the fourth hospital day, the parotid swelling had resolved on the left side, but swelling was still present on the right, and had progressed from being soft to firm on palpation. Noting our frequent examinations, the mother volunteered her impression that the right side of the child's face had seemed to be larger than the left since birth.

Because of the physical examination and the additional history, a computerized tomographic (CT) scan of the facial bones was obtained. The CT scan showed an abnormal right mandible. The mandible was described as irregular and "moth-eaten" in appearance, with diffuse enlargement, suggesting a permeative process. The differential was given as osteomyelitis, which was considered unlikely because of the sharp margins, hemangioma, and histiocytosis, or Letterer-Siwe disease. A bone scan, which

Submitted, revised, December 15, 1995.

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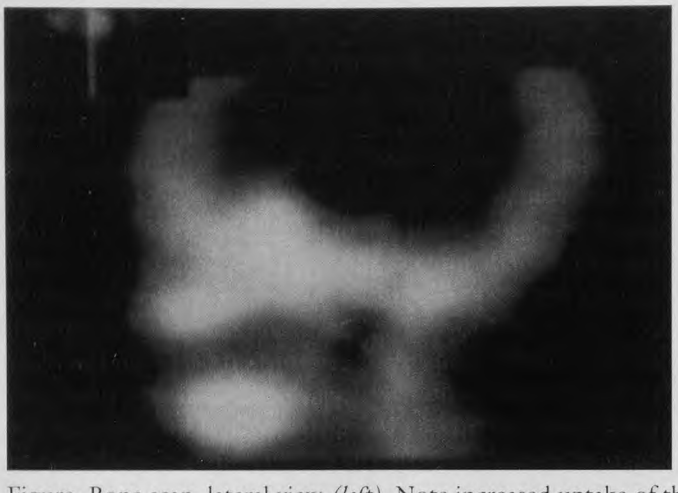
ISSN 0094-3509



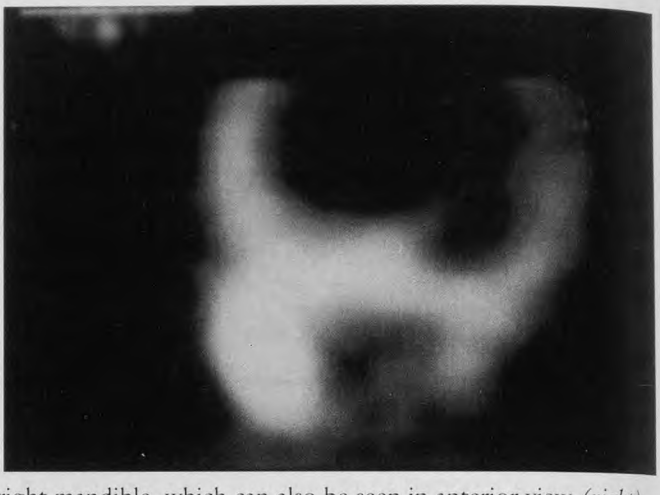


Figure. Bone scan, lateral view *(left)*. Note increased uptake of the right mandible, which can also be seen in anterior view *(right)*.

was obtained to assess the possibility of disease in multiple bones, showed increased uptake in the right mandible only (Figure, left and right). Since no diagnosis was made, a diagnostic bone biopsy was performed.

The child continued to have extensive thrush, despite nystatin therapy, and continued to lose weight daily while in the hospital. The thrush was treated with gentian violet, and a failure-to-thrive workup was done, including T cell and B lymphocyte profiles to assess for an immunocompromised state. The child had been given 24-cal/oz formula with polycose added, yet the weight continued to decline. A nutrition consultation was obtained for feeding recommendations. A calorie count demonstrated that the child was receiving inadequate calories and an intensive oral feeding program was started. Finally, the child's weight stabilized.

Five days following biopsy, the laboratory reported that the biopsy was consistent with Caffey's disease, which usually presents in infancy, and is characterized by fever, soft tissue swelling, and new bone formation that can result in bony deformity. The condition also frequently involves facial bones, resulting in poor feeding and failure to thrive.

Discussion

Infantile cortical hyperostosis was introduced initially in the medical literature in 1939 by Roske[1] and in 1945 by Caffey and Silverman.[2] The disease primarily affects the bones, with monostotic or polyostotic bone compromise, and always includes inflammation. Some body areas, such as the clavicle and the mandible, are affected more often; however, bone involvement can take place almost anywhere in the skeleton.[1–5,10,11] Children are usually affected, with a peak incidence within the first 6 months of life. The diagnosis of Caffey's disease is especially difficult during its initial stages because of the nonspecific nature of the symptoms, which can include fever, poor appetite, malaise, and the nonspecific laboratory abnormalities of leukocytosis, thrombocytopenia, and elevated ESR. The characteristic neoformation of bone and radiographic changes may not occur at the initial onset of symptoms but usually develop within the first 15 to 20 days of the disease.[1–5,10] The differential diagnosis includes a variety of entities, such as trauma, fracture, bacterial osteomyelitis, and neoplasm. The diagnosis relies on exclusion of these more common conditions. The origin of this disease is unknown. Although prevalent from 1940 to 1960, Caffey's disease has since inexplicably declined in prevalence. Hypoxia, local necrosis, and periosteal reaction occur, but what triggers these changes remains a mystery. There are reports in the literature linking the disease to genetic factors, with an autosomal dominant pattern of transmission with variable penetrance.[6] Silverman[7] has proposed a viral cause. Another possibility is that Caffey's disease is immunologic in origin.

The declining incidence of Caffey's disease may be related to the absence of radiographic findings in symptomatic patients or the relatively late presentation of radiographic changes in the course of the disease. Radiographic changes may be present in the absence of constitutional symptoms, and, contrariwise, patients with florid symptomatology might not have radiographic findings on initial presentation.

Caffey's disease usually remits spontaneously, and thus requires no therapy. Particularly severe cases, however, have been treated with prednisone with some success. The disease can cause enough bony deformity that later surgical correction may be required, and the disease can recur.

In the present case, the patient had a diagnostic workup that included bone biopsy and was treated for possible sepsis. The septic workup was negative; the diagnosis was made because of the results of the bone biopsy. At one time this disease was diagnosed on the basis of history, physical examination, and radiographic findings. Ultimately, the patient's fever, physical findings, and failure to thrive can all be explained by a diagnosis of Caffey's disease.

This once-prevalent disease should be part of any differential diagnosis in infants who present with facial bone abnormalities, fever, and leukocytosis. Awareness of this rare disease may permit earlier recognition and a less intensive diagnostic workup.

References

1. Roske G. Eine Eigenartige Knochenerkrankung im sauglingsalter. Monatsschr Kinderheilkd 1930; 47:385–400.
2. Caffey J, Silverman WA. Infantile cortical hyperostosis: preliminary report on a new syndrome. AJR 1945; 54:1–16.
3. Juniper RP. Caffey's disease. Br J Oral Surg 1982; 20:281–7.
4. Marshall GS, Edwards KM, Wadlington WR. Sporadic congenital Caffey's disease. Clin Pediatr 1987; 26(4):177–80.
5. Jakubikova J, Miroslav M, Edita H. Swelling over the face: infantile cortical hyperostosis. Int J Pediatr Otorhinolaryngol 1994; 29: 147–50.
6. Saul RA, Lee WH, Stevenson RE. Caffey's disease revisited: further evidence for autosomal dominant inheritance with incomplete penetrance. Am J Dis Child 1982; 136:56–69.
7. Silverman F. Virus diseases of the bone: do they exist? The Neuhauser lecture. AJR 1976; 126:677–703.
8. Tien R, Barron BJ, Dhene RD. Caffey's disease: nuclear medicine and radiological correlation, a case of mistaken identity. Clin Nucl Med 1988; 13(8):583–5.
9. Newberg AH, Tampas JP. Familial cortical hyperostosis: an update. AJR 1981; 137:93–6.
10. Zacharides N, et al: Infantile cortical hyperostosis. Report of two cases. J Oral Maxilofac Surg 1986; 44:644–8.
11. Behrman RE, Kliegman RM, Nelson WE, Vaughn VC, eds. Nelson textbook of pediatrics. 14th ed. Philadelphia, Pa: WB Saunders Company, 1992.