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Sandra K. Fernbach, M.D.
Department of Radiology, Children’s Memorial Hospital and Northwestern University Medical School, Chicago, Illinois, USA

Radiological Studies

Fig. 1. A lateral roentenogram of the frontal half of skull including the area of the paranasal sinuses shows marked expansion in the area of the upper half of the facial bones, particularly relating to the maxillary sinuses, with a hazy, uniform increase in density overlying the usually aerated paranasal sinuses. In addition, observe the mottling of the frontal bone with spiculation extending from the outer margin of the calvaria into the soft tissues. Some osteopenia is also present in this area of the frontal bones.

Fig. 2. A lateral tomogram centered over the area of the facial bones illustrates once again the striking changes in the area of the facial bones, particularly relating to the usually well-aerated paranasal maxillary antra.

Fig. 3. A frontal plane roentgenogram (Waters position) shows marked expansion and opacification of the facial bones bilaterally, particularly in the area of the antra. The frontal sinuses are well aerated.

Clinical Information

This 23-year-old woman was admitted because of nasal “stuffiness” and excessive lacrimation. Standard views and tomograms of the facial bones were obtained, prior to surgical intervention, which was planned to relieve the nasal obstruction and the lacrimal duct abnormality (Figs. 1–3). Further history is withheld.

Address reprints requests to: Sandra K. Fernbach, M.D., Department of Radiology, Children’s Memorial Hospital, 2300 Children’s Plaza Chicago, IL 60614, USA

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Diagnosis: Beta Thalassemia Affecting the Facial Bones and Skull (Intermediate Form)

The differential diagnosis should include any severe chronic anemia, (e.g. sickle cell anemia, iron-deficiency anemia, the less common hemolytic anemias). Lipid disorders may cause almost similar radiological changes.

The patient was diagnosed as having thalassemia intermedia at the age of six years and had been closely followed since that time, with little need for medical intervention.

Radiological studies of the chest showed a paraspinal soft tissue mass in the retrocardiac region, indicating paraspinal extramedullary hematopoiesis. Expansion of most of the ribs was also present.

Discussion

Beta thalassemia is a chronic hemolytic anemia caused by the inadequate production or total absence of qualitatively normal beta globin chains.

Hemoglobin is a tetramer formed by the binding of two alpha and two beta chains. When insufficient beta chains are present, the alpha chains precipitate out of solution, creating small hypochromic, peculiarly-shaped red blood cells known as leptocytes or target cells [1]. As these abnormal cells are removed from circulation, hyperbilirubinemia and splenomegaly develop.

First described by Cooley and Lee in 1925, beta thalassemia has a relatively high incidence in people of Mediterranean origin. Also known as Cooley anemia, this disorder is inherited as an autosomal recessive. The heterozygote, usually asymptomatic, is the form of this disorder known as thalassemia minor. The homozygote suffers the most severe form of thalassemia (thalassemia major), and generally requires blood transfusion therapy. A third form designated as thalassemia intermedia, is observed in homozygotes who are able to maintain the hematocrit level up to a physiologically tolerable point without transfusion. Individuals with thalassemia intermedia manifest multiple stigmata of the disease, but have a better prognosis than the usual typical homozygote.

Clinically, the typical patient has bronzed skin, mongoloid or rodent facies, and moderate to marked hepatosplenomegaly. The reason for the rodent facies is clearly observed in the radiological studies of the facial bones illustrated in this patient (Figs. 1–3). Digital clubbing of unknown etiology is present to parallel the other clinical signs as well as the abnormal levels of serum transaminases (SGOT and SGPT).

Many radiological features in the patients with thalassemia major are due to enlargement of the marrow space in an attempt to compensate for chronic hemolysis. Roentgenograms of the skull demonstrate widening of the diplic space especially in the frontal and posterior parietal regions. Spicule formation produces the “hair-on-end” appearance described classically in thalassemia, but actually present in only about 5% of patients [2]. Bony overgrowth will cause frontal bossing, maxillary prominence and poorly pneumatized paranasal sinuses and mastoid air cells.

In children, the metacarpals are rectangular or even biconvex. Osteoporosis, cortical thinning and a prominent trabecular pattern lends a honeycomb appearance to the medullary shafts in these and most other tubular bones. As the patient matures and the red marrow compartment centralizes, the changes in the hands and distal extremities regress. At this time the abnormal features in the spine (osteoporosis and prominent trabeculae) worsen. Other bony changes include: (1) delayed appearance and early fusion of ossification centers which are responsible for dwarfism and deformity; (2) bowing of the long bones; (3) and rarely, ischemic necrosis of the femoral head [3].

Various visceral changes are also present, including enlargement of the liver, spleen or kidneys which may be appreciated on standard abdominal roentgenograms [4]. Extramedullary hematopoiesis may also be observed, manifesting as paraspinal masses (present in this patient) or soft tissue masses at the ends of the ribs. The hemolytic process causes changes in the metabolism of the bile and radiopaque calcium bilirubinate gallstones are often produced. When hemochromatosis develops secondary to multiple transfusions, computed tomographic scanning will demonstrate that the Hounsfield number of the hepatic parenchyma is elevated, paralleling the amount of iron present in the liver tissue. Houang et al. suggest using computed tomography rather than biopsy to follow the course of the involvement of the liver [5].

Otolaryngologists are aware of enlargement of the adenoids and tonsils, hypertrophy of the nasal turbinates and hearing deficits associated with changes in the bones of the middle ear [6]. Soft tissue involvement can also extend to the peripher-