Consultant360 Multidisciplinary Medical Information Network

WHAT'S YOUR DIAGNOSIS?

PEER REVIEWED

What Is Causing an Infant's Facial Swelling?

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A healthy 55-day-old full-term infant presented to the emergency department (ED) with a 4-day history of left-sided facial swelling with increasing firmness (**Figure 1**).

The parents initially had been concerned about a possible allergic reaction to the infant's formula and had switched him to goat's milk without improvement. The patient then had been seen by his pediatrician, who recommended evaluation in the ED. The parents denied fever, upper respiratory tract symptoms, decreased oral intake, or difficulty breathing in the child.

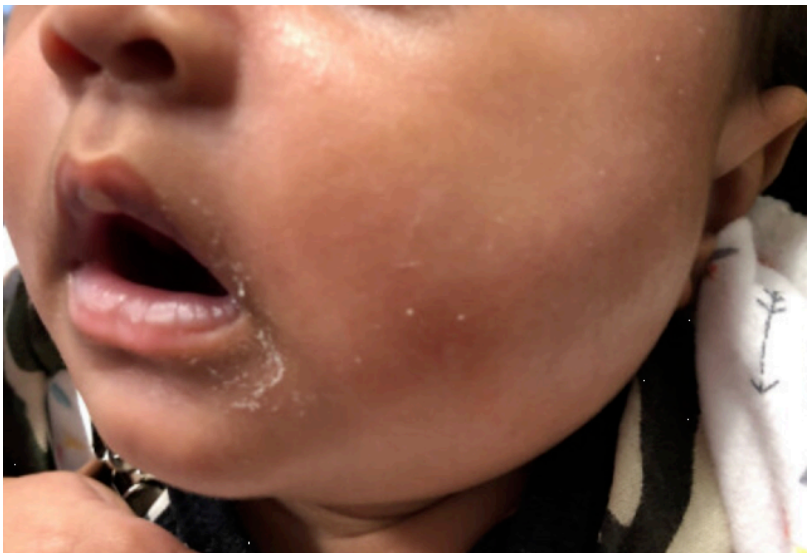


Figure 1. *A 55-day-old infant presented with a 4-day history of left-sided facial swelling with increasing firmness.*

In the ED, ultrasonography of the neck was ordered, the results of which demonstrated an asymmetric enlargement of the left masseter muscle (**Figure 2**).

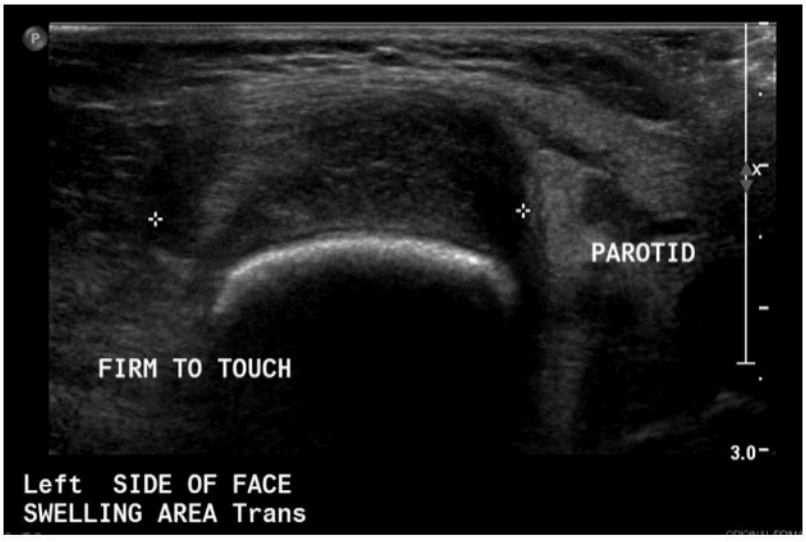
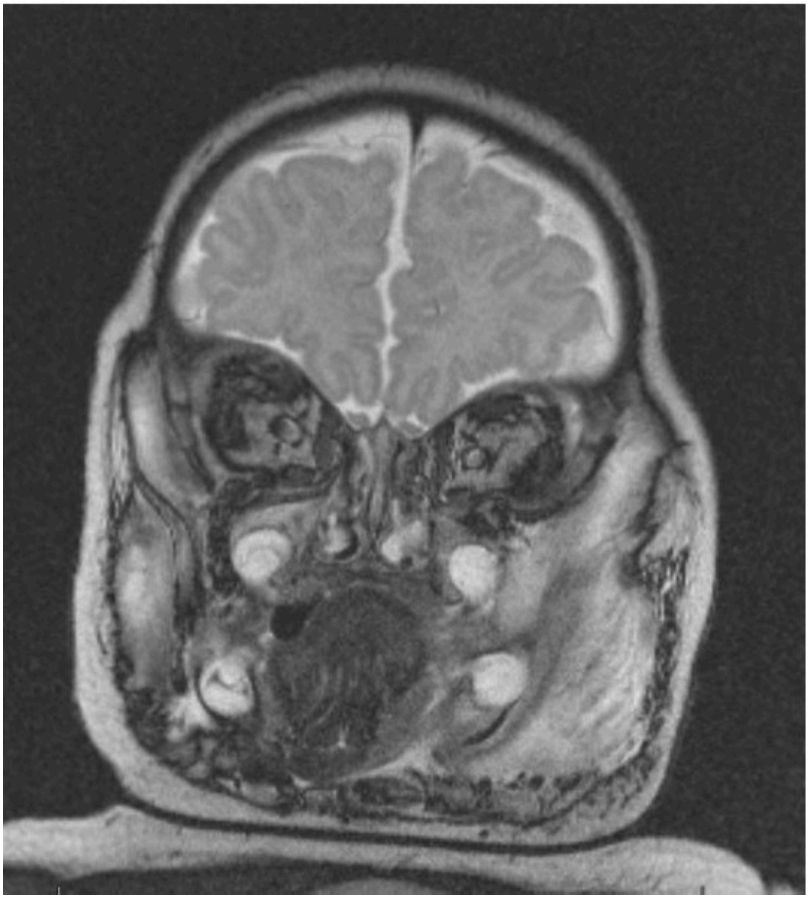


Figure 2. *Ultrasonography demonstrated an asymmetric enlargement of the left masseter muscle.*

While in the ED, the patient had a temperature of 38.0°C. A full sepsis workup was initiated with blood, urine, and cerebrospinal fluid (CSF) cultures, and the patient was started on empiric ceftriaxone.

Results of a complete blood cell count were significant for leukocytosis, with 23,500 white blood cells/ μL with 37% neutrophils, 45% lymphocytes, and 14.8% monocytes. The C-reactive protein (CRP) level was elevated at 3.94 mg/L, and the erythrocyte sedimentation rate (ESR) was elevated at 89 mm/h. Results of a comprehensive metabolic panel, CSF indexes, and urinalysis were unremarkable. The patient was treated with empiric ceftriaxone, and an otorhinolaryngologist was consulted, who recommended magnetic resonance imaging (MRI) for further evaluation (**Figure 3**).



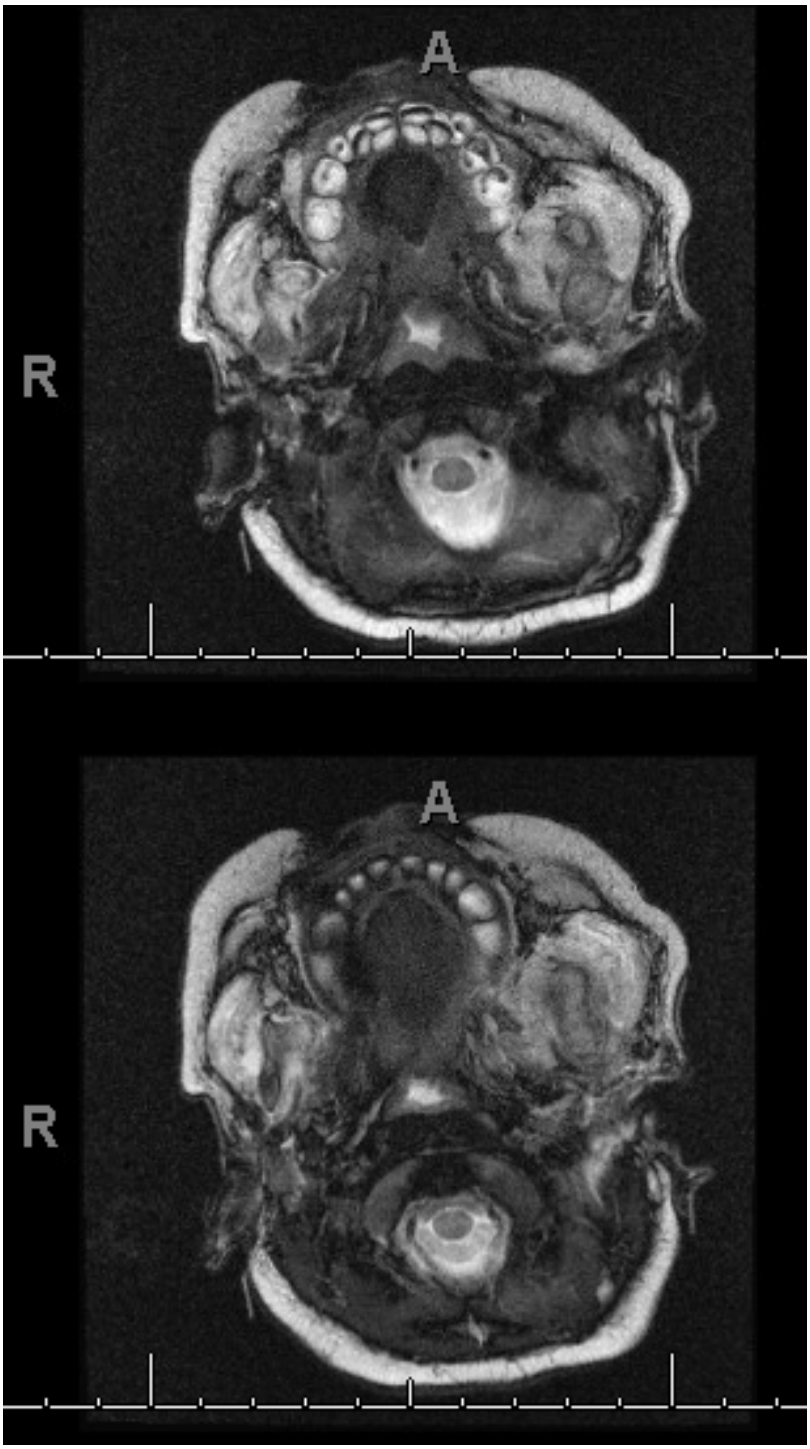


Figure 3. *MRI scans of the head and neck.*

What is the cause of this patient's facial swelling?

A. Rhabdomyosarcoma

B. Osteoid osteoma

C. Cold panniculitis

D. Cellulitis

E. Infantile cortical hyperostosis

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Answer: Infantile Cortical Hyperostosis

Infantile cortical hyperostosis, or Caffey disease, is a rare and generally self-limiting bone disorder characterized by fever, hyperirritability, and new periosteal bone formation leading to cortical thickening (hyperostosis) of the affected bone or bones and swelling of overlying soft tissue. Its mean age of onset is 9 weeks, and it usually regresses spontaneously by 2 years of age.1

Although it may occur in many locations, including the mandible, clavicle, scapula, skull, ilium, and long bones, the mandible is most frequently affected (90% of cases),2 and its involvement is nearly pathognomonic.3 Understanding of mandibular involvement in infantile cortical hyperostosis is useful in differentiating the condition from child abuse, since periosteal reaction of a single bone (except the mandible) is often suggestive of trauma.

The pathogenesis of infantile cortical hyperostosis has not been completely elucidated, although familial studies have described a missense mutation on 17q21 affecting the gene encoding the α1 chain of type I collagen present in bone, skin, ligaments and teeth.3

Imaging studies are essential in making the diagnosis of infantile cortical hyperostosis. Plain radiographs show periosteal bone growth with thickened cortices.4 MRI findings can sometimes precede plain-film findings and include thickening of the periosteum with edema and inflammation of the adjacent soft tissues.3 Laboratory findings include elevated inflammatory markers ESR and CRP, as well as elevated levels of alkaline phosphatase and immunoglobulins.1

DIFFERENTIAL DIAGNOSIS

Rhabdomyosarcoma is a malignant skeletal muscle neoplasm and is the most common soft tissue tumor in children. Although a rhabdomyosarcoma can occur anywhere in the body, the

most common sites are the head and neck (approximately 40%), genitourinary tract, and the extremities.5 The majority of rhabdomyosarcomas are sporadic; however, the condition can be associated with Li-Fraumeni syndrome, Costello syndrome, Beckwith-Wiedemann syndrome, and Noonan syndrome.5 Diagnosis is made with biopsy, immunohistochemistry, and genetic testing. Patients often present with a painless, infiltrative, rapidly expanding mass, with or without overlying skin changes.6

Osteoid osteoma is a benign tumor of the bones characterized by a small radiolucent nidus.7 It typically presents during the second and third decade of life, with the majority of cases (approximately 80%) occurring in the long bones, especially the proximal femur. However, cases of mandibular involvement have been reported.8 The most common symptom is progressively increasing pain, which is often worse at night.

Cold panniculitis is caused by lowered skin temperatures leading to crystallization in the subcutaneous fat, followed by an inflammatory reaction. Due to the higher ratio of saturated to unsaturated fatty acids in infants, and the higher melting point than the subcutaneous fats of older children, this is almost exclusively seen in infants and young children. It is clinically characterized by erythematous indurated nodules appearing 1 to 2 days after exposure to cold.9

Cellulitis is the result of a bacterial infection of the deeper dermis and subcutaneous fat. It manifests clinically as an acute, spreading, poorly demarcated area of skin erythema, warmth, swelling, and tenderness. It is nearly always unilateral. Fever and other systemic manifestations, such as malaise and leukocytosis, may also be present. In immunocompetent patients, it is usually thought to be caused by group A streptococci, with *Staphylococcus aureus* being a significant but less common cause.10

DIAGNOSIS AND TREATMENT

After admission, brain MRI was obtained, the results of which demonstrated diffuse edema and enhancement involving the bilateral muscles of mastication and overlying soft tissues (left greater than right) with abnormal signal and periostitis involving the left hemimandible, consistent with infantile cortical hyperostosis. A skeletal survey was obtained, the results of which showed no other areas of hyperostosis.

When it was determined that the etiology was no longer infectious, the antibiotics were discontinued, and the patient remained afebrile. The patient was well appearing, afebrile, and tolerating oral feedings well, and he was discharged home with scheduled follow-up with genetics and orthopedics specialists. The patient continued to do well with no additional problems, and the family received genetic and prognosis counseling, with genetic testing pending.

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