A 4-year-old Boy With Fever

Joel Charrow, MD

An 4-year-old boy presents with fever. The mother reports that he has had multiple ear infections in the past, is “always congested,” and has always been small. Everyone else in the family is of normal stature. They have just relocated to the area from a small farming community. No prior medical records are available.

On examination, the boy’s oral temperature is 39 degrees C, and the left tympanic membrane is bulging and red. He appears short and has a relatively large head; his height is far below the 5th percentile, and his head circumference is above the 95th percentile (Figure 1). He has an accentuated lumbar lordosis (Figure 2). His limbs appear relatively shorter than his trunk.

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Editor's note: Each month, this department features a discussion of an unusual diagnosis in genetics, radiology, or dermatology. A description and images are presented, with the diagnosis and an explanation of how the diagnosis was determined following. As always, your comments are welcome.
DISCUSSION

Although it was acute otitis media that brought this child to medical attention, the presence of marked short stature (ie, dwarfism) is highly suggestive of a skeletal dysplasia, especially when accompanied by disproportion of the limbs and trunk. Specific diagnosis requires radiographic examination of the bones. An x-ray skeletal survey should be obtained, including all of the long bones, pelvis, two views of the spine and skull, and antero-posterior views of the hands and feet.

This boy’s physical features are highly suggestive of achondroplasia, the most common bone dysplasia. These include short stature (with rhizomelic shortening of the limbs), trident hand (Figure 3), accentuated thoracic kyphosis and lumbar lordosis, and macrocephaly. The base of the skull is shortened, resulting in a small nasopharynx and eustachian tube dysfunction. Chronic congestion, mouth breathing, and snoring are common. Intelligence is normal.

The diagnosis is confirmed by the x-ray findings. There is progressive narrowing of the interpediculate distance in the lumbar spine, instead of the normal increase in this distance.
in the lumbar region (Figure 4, see page 520). There is also a characteristic shape of the pelvis, with a very small lower ileal segment ("paddle without a handle") and a narrow sacrosciatic notch (Figure 5).

In infancy and early childhood, the foramen magnum is foreshortened and may cause compression of the upper cervical spinal cord. In severe cases, this may cause central apnea and sudden death. The abnormal configuration of the nasopharynx may result in partial upper airway obstruction, obstructive sleep apnea, cor pulmonale, and right heart failure. Later in life, spinal stenosis may result in myelopathy in lower spinal cord segments.

The macrocephaly is often confused with hydrocephalus. Although the ventricles may appear slightly enlarged on neuroimaging studies, true hydrocephalus is rare. Prior fossa decompression is necessary in some cases to relieve cord compression related to the stenosis of the foramen magnum.

This disorder results from mutations in the fibroblast growth factor 3 (FGFR3) gene, with more than 95% of cases related to a substitution of arginine for glycine at amino acid position 380. Although the condition is inherited as an autosomal dominant trait with complete penetrance, more than 85% of affected children are born to normal parents and are the result of new mutations.

Dr. Charrow is professor of pediatrics, Feinberg School of Medicine, Northwestern University, Chicago, IL, and head of the Section of Clinical Genetics, Children’s Memorial Hospital, Chicago.