Misdiagnosis in a Child Presenting with Bulging Anterior Fontanel: Differential Diagnosis

Ön Fontanel Kabarıklığı ile Başvuran Çocuk Hastada Tanısal Karışıklık

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ABSTRACT Fontanel bulging is an important clue in the diagnosis of meningitis. Subdural hematoma, bleeding secondary to whiplash or shaking baby, intracranial tumors, human herpes virus infection, maple syrup urine disease, urea cycle enzyme defects, vitamin A poisoning, pseudotumour cerebri, hypophosphatasia and diphtheria-pertussis-tetanus vaccination are other causes for fontanel bulging. We reported a 6-month-old infant with osteopetrosis who had been treated with antibiotics due to presumptive diagnosis of meningitis because of marked fontanel bulging associated with fever. We aimed to remind physicians to be aware of causes of fontanel bulging other than meningitis to avoid misdiagnosis and its untoward consequences.

Key Words: Osteopetrosis, meningitis

ÖZET Fontanel kabarıklığı menenjit tanısında önemli bir ipucudur. Subdural hematom, bebeğin sarsılması ya da silkelenmesi sonucu kanama, intrakraniyal tümörler, insan herpes virüsü enfeksiyonu, akçaağaç şurubu idrar hastalığı, üre döngüsü enzim kusurları, A vitamini entoksikasyonu, hipofosfatazya, psödotümör serebri, difteri-boğmaca-tetanoz aşılaması da fontanel kabarıklığını oluşturan diğer nedenlerdendir. Biz burada, ateşin eşlik ettiği belirgin fontanel kabarıklığı nedeni ile menenjit düşünülerek antibiyotik tedavisi verilen 6 aylık osteopetrozis olgusunu sunduk. Yanlış tanının ve buna bağlı istenmeyen sonuçların önlenmesi için klinisyenlere fontanel kabarıklığının menenjit dışı diğer nedenlerinin tanınmasında dikkatli olmaları gerektiğini tekrar hatırlatmayı amaçladık.

Anahtar Kelimeler: Osteopetrozis, menenjit

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Fontanel bulging acts as a safety valve in case of increased intracranial pressure. It is an important clue in the diagnosis of meningitis and may be the only early physical sign of meningitis in an infant.[1] Several other causes of fontanel bulging include subdural hematoma, bleeding secondary to whiplash or shaking baby, intracranial tumors, human herpes virus infection, hypophosphatasia, maple syrup urine disease, urea cycle enzyme defects, vitamin A poisoning and diphtheria-pertussis-tetanus vaccination.[1-3]

We presented a 6 months old infant with osteopetrosis with a presumptive diagnosis of meningitis due to marked fontanel bulging associated with fever. We aimed to remind that physicians to be aware of the causes of bulging anterior fontanel to avoid misdiagnosis and its consequences.

CASE REPORT

The patient was a 6-month-old girl who had been treated with intravenous antibiotics with a presumptive diagnosis of meningitis because of marked fontanel bulging associated with fever without performing lumbar puncture in a primary care hospital. She was referred to our hospital for further evaluation. She was the first child of healthy consanguineous parents. Her previous health history was not remarkable. On admission to our hospital, her vital signs were all normal. Her head circumference was 38.5 (25 percentile), height 61 cm (50 percentile); and weight 5.2 kg (10-25 percentile). Her physical examination revealed wide anterior fontanel (3 x 4 cm) and hepatosplenomegaly with the liver 2 cm and the spleen 1 to 2 cm below the costal margin (Figure 1). Additionally, proptosis and optic nerve pallor was observed on ophtalmologic examination of the patient. The rest of her physical examination was normal. On admission to our hospital, her laboratory evaluation was as follows: Hemoglobin 10.2 g/dL, platelet count 347 x 10^9/L , white blood cell count 17.9 x 10^9/L with differential count of 20% polymorphonuclear leucocyte, 64% lymphocyte, 6% monocyte, 4% normoblast, 4% promyelocyte, 2% myelocyte which was consistent with leukoerytroblastic reaction. Biochemical tests were all normal except hypophosphatemia. The phosphorus level was 3.5 mg/dL (normal, 5-10.8 mg/dL). 25-hydroxy vitamin D level was elevated at 220 ng/mL (normal, 7.5-75 ng/mL), and parathormone level was high at 440 pg/mL (normal, 11-68 pg/mL). She underwent a skeletal survey and had the evidence of bone sclerosis throughout the skeleton, the presence of "bone within a bone" appearance and metaphyseal irregularity of tubular bones. Computerized tomography (CT) scan of the head demonstrated bulging of the brain from the anterior fontanel and narrowing of the orbital fissure and there was no ventriculomegaly (Figure 2). Three-dimensional CT of the head showed the bony fusion of the coronal sutures and the narrowing of the lambdoid and sagittal sutures. Visually evoked potential (VEP) showed slightly delayed flash responses. The patient was diagnosed as osteopetrosis and was sent to an advanced center where pediatric neurologist, ophtalmologist and otorhinolaryngologist could be involved in the periodic follow-up of the patient and bone marrow transplantation could be carried out.

DISCUSSION

In osteopetrosis, defective osteoclastic function results in bone thickening. It causes thickening of the skull, lacking of intracranial volume for expansion and impaired outflow of blood of venous sinuses at the exit of foramina which lead to elevation of intracranial pressure and result in papilledema and fontanel bulge.[4] The presence of proptosis, fontanel bulging and optic canal stenosis on CT scan were all related with neurological consequences of osteopetrosis in our patient. The association of fever with fontanel bulging and the false concerns as

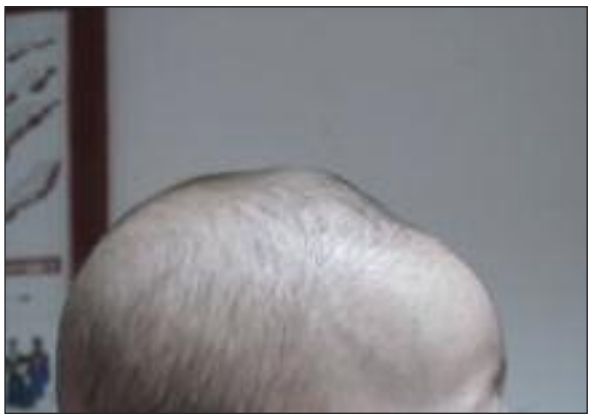

FIGURE 1: Image of fontanel bulging.

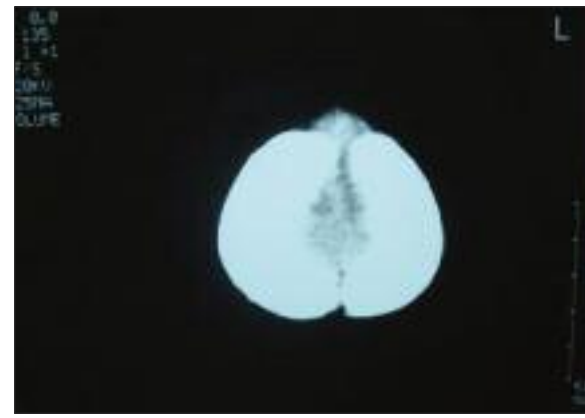

FIGURE 2: Bulging of the brain from anterior fontanel on axial CT scan.

to whether the bulging anterior fontanel was predictive of meningitis, might have caused confusion, and contribute to the misdiagnosis of meningitis in our patient. If imaging were available, it would be standard practice to image this child's head following initial evaluation. If imaging were not readily available, it would be malpractice not to treat this child with antibiotics for presumptive meningitis initially. The consequences of missing meningitis are very severe.

Compensatory extramedullary hematopoiesis with resultant leukoerytroblastic reaction is the hematologic characteristic of osteopetrosis that consist of circulating normoblasts, teardrop shaped poikilocytosis, early myelocytes and hepatosplenomegaly as in our patient.[5]

In the literature rickets with hypophosphatasia and nutritional rickets are mentioned as the causes of fontanel bulging due to delayed bone growth in the presence of normal brain growth.[2] Hypophosphatemia, elevated levels of serum parathormone and 25-hydroxy vitamin D and metaphyseal irregularity of tubular bones may be the signs of developing rickets in our patient, which is a common and paradoxical feature of osteopetrosis.[6] This might have contributed to the exaggeration of fontanel bulging in our patient by the mechanism mentioned in the literature.[2]

CONCLUSION

This case provides an important lesson on the evaluation of any patient with fontanel bulging that physicians should be careful in recognition of causes of fontanel bulging other than meningitis thus avoiding possible misdiagnosis and untoward consequences. We suggest all physicians to keep osteopetrosis in mind as another cause of fontanel bulging.

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