Persistent open anterior fontanelle in a healthy 32-month-old boy

HILDA M. DE GAETANO, DO
JOSEPH S. DE GAETANO, DO

Delayed closure of the anterior fontanelle is often associated with significant disease entities. Range of normal closure of the anterior fontanelle is 4 to 26 months. Increased intracranial pressure, hypothyroidism, and skeletal anomalies are common etiologic factors. History, physical examination, and diagnostic testing rule out most disorders. Once these disorders have been ruled out, it is important for the physician to realize that a persistent open anterior fontanelle beyond the accepted ranges of closure can be a normal outlier.

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Well-child care is an important aspect of pediatrics and family medicine. Each day, physicians do physical examinations on thousands of children worldwide. As we know through our medical education and clinical experience, children attain many physical milestones throughout their development. Physicians use these physical milestones to assess the health and well-being of their young patients and regard variations from normal accepted ranges as red flags that can indicate a disease process.

Palpation of the anterior fontanelle is an integral part of any well-child examination. Much information can be gained from this simple maneuver. Closure of the anterior fontanelle is an important physical milestone that indicates a child’s well-being. The range of normal closure is between 4 and 26 months.1 Delayed closure of the anterior fontanelle can be associated with many disease processes; however, careful review of the literature reveals that once these specific disease processes have been ruled out, delayed closure can be a normal finding.1 The purpose of this report is to familiarize physicians with those etiologic factors commonly associated with delayed closure of the anterior fontanelle. A review of the workup to rule out these entities will be provided, and we will show that once common diseases have been ruled out, a persistent open anterior fontanelle can be a normal outlier.

S.D. is a 32-month-old African American boy who presented to our clinic for a well-child physical examination. He had no specific complaints at the time of presentation, and his examination was essentially unremarkable except for the finding of an open anterior fontanelle measuring 3 cm × 1.5 cm. This was noted to be an unusual finding considering the patient’s age.

S.D. was born to a 21-year-old gravida II para I African American woman at term gestation. His mother’s prenatal course had been uncomplicated. Similarly, her previous pregnancy was without complication. At birth, S.D. weighed 3019 g, was 49 cm long, and had a head circumference of 36 cm—all appropriate for gestational age. Routine plotting on a standard growth curve chart for newborn males revealed that he was at the 50th percentile for weight and head circumference, and at the 25th percentile for height. Physical examinations done at the time of birth and at discharge from the hospital all were within normal limits. Routine newborn laboratory work including hematocrit, glucose, total bilirubin, and metabolic screen also was unremarkable. S.D. had no known family history of any metabolic, endocrine, or dysmorphogenic diseases.

S.D. continued to routinely follow up in our clinic. He received all of his immunizations at the appropriate intervals as well as a screening hematocrit and lead level at 1 year. His medical history is only remarkable for several bouts of bronchiolitis, periodic exacerbations of reactive airway disease, and one hospitalization at 6 months for pneumonia.

Throughout the course of his well-child examinations, S.D.’s head circumference continued to plot at the 50th percentile, his weight remained at the 50th percentile, and his height at the 25th percentile. He has always met all milestones for motor, language, and social skills per routine Denver developmental screening. He has never had any dysmorphic features, nor has he been exposed to drugs or toxins.

At S.D.’s initial visit, the fontanelle was neither bulging nor pulsatile. As this was a peculiar finding in a 32-month-old child, a simple skull series was obtained to rule out underlying pathology. The skull films depicted the open fontanelle as well as possible diastasis of the coronal sutures. A computed tomographic (CT) scan of the head was recommended to rule out causes of increased intracranial pressure.

S.D. was admitted to the hospital for sedation before the head CT scan to obtain an optimal study. Laboratory
work to include a screening urinalysis, complete metabolic profile, complete blood count, and thyrotropin was ordered in an effort to rule out metabolic as well as endocrine etiologic factors. The CT scan revealed the open fontanelle; however, no pathologic intracranial processes were noted. Simultaneously, all laboratory work returned with normal values. Potentially deleterious causes of persistent open anterior fontanelle were ruled out. S.D. was discharged after his parents were reassured that his physical examination finding was a variant of normal and that closure would eventually occur.

Persistent open anterior fontanelle can be a common finding in children. This normal variant must be recognized, given the vast number of well-child examinations done annually.

In 1949, Milton Aisenson, MD, reviewed the records of two child health stations of the New York City Department of Health for over 10 years to determine the normal age at which the anterior fontanelle closes. Examinations of 1677 infants were recorded to note the age of closure of the anterior fontanelle. The range of closure was determined to be between 4 and 26 months. Ninety percent of the children’s fontanelles closed between 7 and 19 months, and 42% of the fontanelles closed before 1 year. This study set the guidelines by which we examine our infant patients and counsel their parents about the closure of the anterior fontanelle.

Delayed closure of the anterior fontanelle can be associated with multiple diseases, most of which have dysmorphic features that should facilitate early recognition. Simple radiographic or laboratory studies rule out other common causes. Increased intracranial pressure is the most common cause of delayed closure of the anterior fontanelle. Multiple etiologic factors are responsible for this phenomenon. Hydrocephalus, subdural hematomas, porencephalic cysts, and tumors are most frequently seen. All are easily and routinely identifiable via plain skull series or CT scanning of the head.

Many skeletal disorders are responsible for delayed closure of the anterior fontanelle. Achondroplasia, osteogenesis imperfecta, vitamin D deficiency—rickets, and cleidocranial dysostosis are the most common of these. These diseases present with characteristic physical findings and are confirmed by associated laboratory and x-ray abnormalities.

Chromosomal defects as well as dysmorphogenetic syndromes also predispose infants to delayed closure of the anterior fontanelle. Down, trisomy 13, trisomy 18, Russell-Silver, Rubinstein-Taybi, and Robinow’s syndromes commonly encompass this physical finding within the constellation of findings associated with that particular syndrome. All are routinely identified early in child development as the result of their significant dysmorphic features.

Endocrine disorders as well as drug and toxin exposure are also associated with delayed closure of the anterior fontanelle. Commonly, hypothyroidism, fetal hydantoin syndrome, aminopterin-induced malformations, and aluminum toxicity can all be associated with a persistent open fontanelle. These also are easily ruled out via a thorough history, blood levels, and thyroid function screening. All of the previously mentioned disease processes had been sufficiently ruled out via the multiple histories, physical examinations, x-rays, and laboratory studies that S.D. had undergone since birth.

The significance of a persistent open anterior fontanelle in an otherwise healthy 32-month-old child remained to be determined.

A search of the literature yielded few results. One source stated that in a healthy child with a persistent open fontanelle who has continued to plot accordingly on growth charts, the finding is considered an outlier with no specific significance. Yet another source stated that as long as a child’s head circumference progresses along a normal curve and the neurologic and ocular fundoscopic examinations yield normal results, no further diagnostic studies are required.

The persistent open fontanelle probably has a familial inheritance pattern associated with it. Closure simply occurs at a time beyond the accepted range of normal. The importance of recognizing the many etiologic factors responsible for a persistent open anterior fontanelle is crucial for physicians who care for children. Physicians must know the multiple syndromes, diseases, and toxic exposures that can cause a delayed closure.

This case report and literature review demonstrate that once simple laboratory studies and x-rays are performed and the previously mentioned maladies are ruled out, a persistent open anterior fontanelle can be a normal finding. This carries tremendous clinical relevance as we counsel our patients’ parents about the ramifications of this discovery. Parents can be told that there is probably a familial inheritance associated with delayed closure of the anterior fontanelle and that with time the fontanelle should close.

References