

CASE STUDIES

Familial Autosomal Dominant Brachydactyly Using 3D Surface Rendering and Multiplanar Reconstruction

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DOI: 10.1177/8756479304263539

This case report demonstrates a normal fetus with familial autosomal dominant brachydactyly. The sonographic examination used 3D surface rendering and multiplanar reconstruction. The mother had a personal as well as family history of the inherited anomaly.

Key words: brachydactyly, autosomal dominant inheritance, 3D imaging

Case Presentation

A patient in her late 30s, gravida 1, para 0, presented at 17 weeks gestation for an anatomical survey. Her personal history and family pedigree were remarkable for autosomal dominant brachydactyly. Both of her hands were affected (Fig. 1). The thumbs were normal, but the second through fifth fingers were compromised. Multiple family members had inherited the anomaly going back three generations. Given her history, there was a 50% probability that the fetus would be affected.

During the initial 2D sonographic examination using a Toshiba Nemio system, with a 5-MHz curved-array transducer, the fetal fingers did appear to be shortened. However, given the small size of the hands at 17 weeks gestation, a follow-up examination was scheduled. The patient returned at 27 weeks gestation. Standard 2D sonography demonstrated what appeared to be shortened digits on both hands (Figs. 2, 3), but this was far from conclusive. Using Biomedicom SonoReal/Sono 3-D software, surface renderings of both hands were attempted. This proved to be very challenging. Successful images required the fetal hands to be temporarily motionless and free of obstructing structures. Persistence, as well as the 3D surface renderings, made the diagnosis undeniable (Figs. 4, 5). Further analysis with multiplanar reconstructions allowed for a very precise evaluation of each



FIG. 1. Maternal hands.

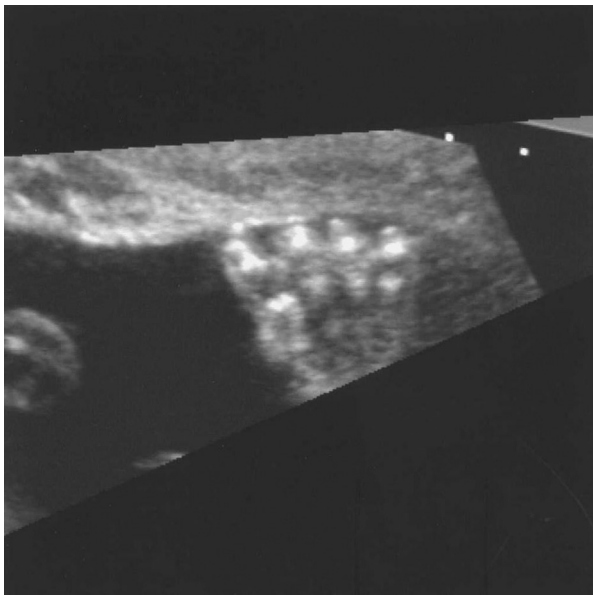


FIG. 2. A 2D image of the fetal left hand at 27 weeks gestation.

individual tuft. Each phalanx was reconstructed in either the coronal or sagittal planes for optimal resolution. All of the distal tufts of the second through fifth fingers were absent or fused with the middle phalanx, and the middle tufts had varying degrees of hypoplasia. Both thumbs were normal.

A male child was delivered at term. There was type A1 brachydactyly involving both hands (Fig. 6). The child was otherwise normal.

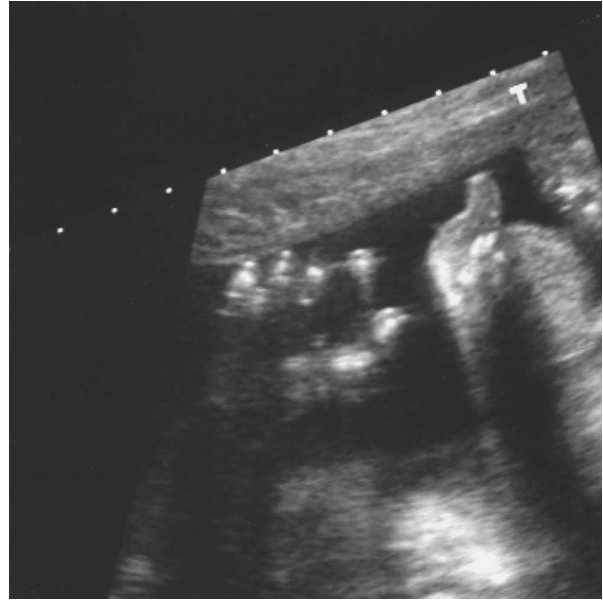


FIG. 3. A 2D image of the fetal right hand at 27 weeks gestation.

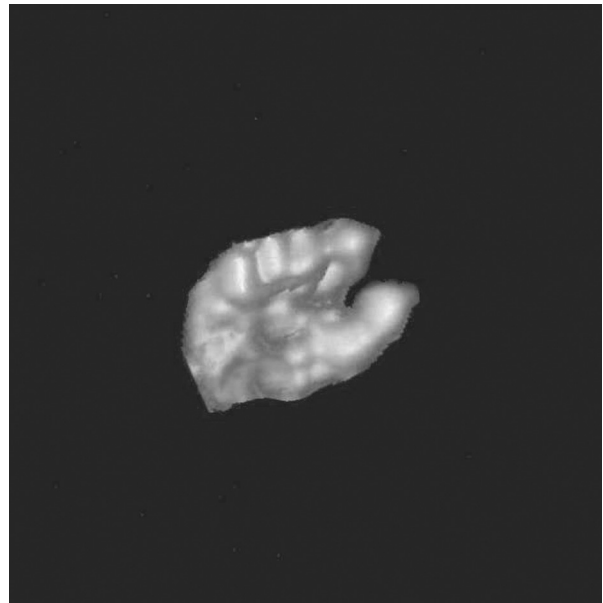


FIG. 4. A 3D surface rendering of the fetal right hand.

Discussion

Brachydactyly is an autosomal dominant disorder in which the affected individuals have short or stubby fingers and toes. During the initial period of

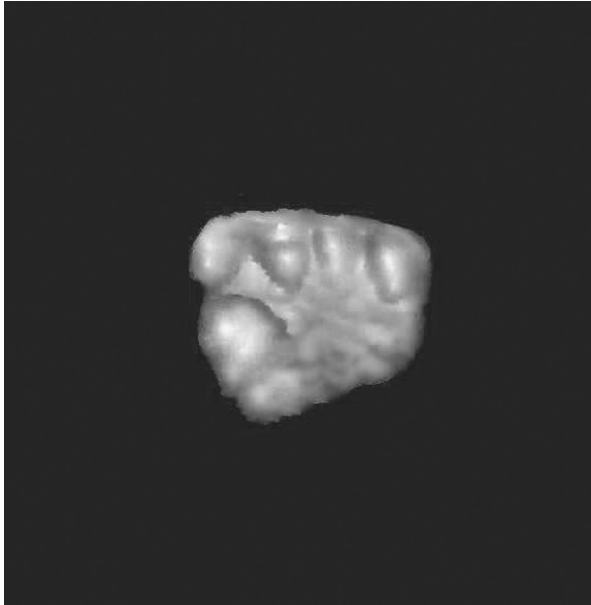


FIG. 5. A 3D surface rendering of the fetal left hand.

bone growth in the phalanges, the hallmark of the anomaly is the development of a “cone epiphysis,” whereby the apex protrudes into the adjacent metaphysis. Epiphyses close prematurely, leading to short, broad tubular bones.¹ Bell² classified brachydactyly into seven types: A1, A2, A3, B, C, D, and E. Although Bell’s classifications have had some modifications over the years, they are still in use today. The type A brachydactylies have the shortening confined mainly to the middle phalanges. In the A1 type, the middle phalanges of all the digits are rudimentary or fused with the terminal phalanges. Brachydactyly type A1 has been shown to be caused by a mutation in the *IHH* gene, located on chromosome 2.³ Several classes of genes have been implicated in the control of distal limb development, including homeobox-containing genes, some members of the homeobox gene family, and genes encoding growth factors.⁴

Autosomal dominant inheritance is marked by the primary feature that one copy of an allele is sufficient for expression of a trait; the gene located on 1 of the 22 autosomes (i.e., not the X or Y chromosome) is expressed in the heterozygous state. Each affected person has at least one affected parent (Fig. 7). An affected person has a 50% chance of passing the trait to a child. Both males and females



FIG. 6. Postnatal photograph of child’s right hand.

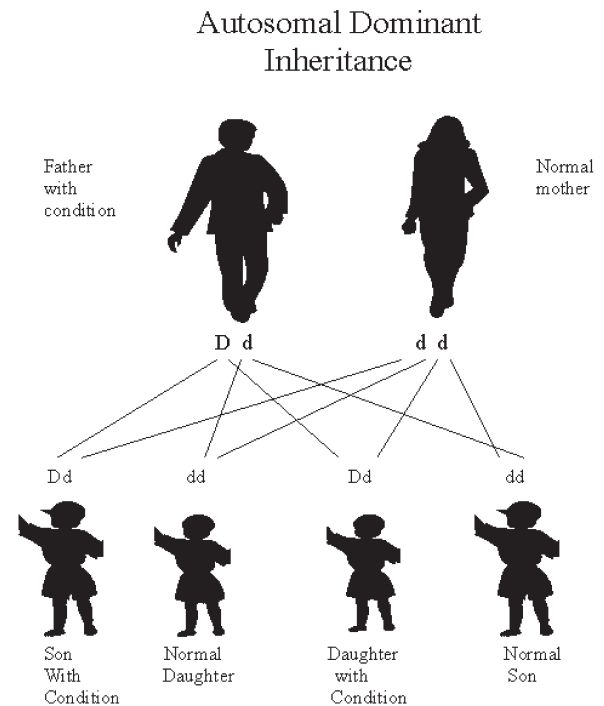


FIG. 7. Autosomal dominant inheritance.

are equally likely to be affected, and two affected people can have an unaffected child. Exceptions may occur if the affected person is the result of a new mutation. Furthermore, if the parent transmitting the gene did not show the trait but carries the allele, this is known as incomplete penetrance—that is, the inconsistent phenotypic expression of a gene even though it is present. In addition, when the parent of the affected individual expresses the

gene but in ways that are not readily recognized, this is known as variable expressivity.⁵ The pattern of pedigree in the family in this case study was typical for inheriting the anomaly.

Comments

The sonographic diagnosis of brachydactyly can be difficult using standard 2D techniques. The addition of 3D surface rendering remarkably improves the ability to make the diagnosis. Multiplanar reconstructions in both the coronal and sagittal planes allow for a better definition of each phalanx for aid in overall classification.

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