Preaxial hand polydactyly in association with lumbar hemivertebra

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DESCRIPTION

A 20-month-old boy presented with a double right thumb. His parents reported no health complaints other than the skeletal disorder involving his right hand. He is the first child born to non-consanguineous healthy parents. His mother is 24 years old. There is no family history of any kind of congenital skeletal abnormalities. No history of drug, smoke, alcohol or radiation exposure during pregnancy is present. The patient was born full-term with normal vaginal delivery. No other significant health problem is present in the history of his infancy. On physical examination, duplication of the right thumb was observed. The thumbs showed parallel alignment with each other. Both feet were normal. Physical examination of the child additionally revealed a slight spinal curvature to the left. Anteroposterior and lateral radiographs of the right hand demonstrated duplication of both the proximal and distal phalanges of the thumb consistent with Wassel type IV polydactyly (figure 1). On anteroposterior spinal radiograph, there was a left convexity in the lumbar spine with a left lateral hemivertebra at L2 level. Spina bifida involving the L5 vertebra was also evident in the spinal radiograph (figure 2).

Preaxial polydactyly is a congenital skeletal disorder that is characterised by the duplication of the thumb. It occurs in 8 per 100.000 births and manifests in different forms depending on the level of the bifurcation. According to the Wassel's classification system: type I describes a bifid distal phalanx; type II, duplicated distal phalanx; type III, bifid proximal phalanx; type IV, duplicated proximal phalanx; type V, bifid metacarpal; type VI, duplicated metacarpal; and type VII, triphalangism.¹ The case we currently present exhibits Wassel type IV, which is the most common form of preaxial polydactyly. Polydactylies have been reported to

Figure 2 Anteroposterior whole spinal (A) and lumbar (B) radiographs show a left convexity in the lumbar spine with a left lateral hemivertebra at L2. Note spina bifida involving L5 vertebra.

rarely associate with other congenital abnormalities except for some established syndromes, including trisomy 13, Down, Bardet-Biedl, Carpenter, and Joubert syndromes. When syndromes are excluded, syndactyly is the most frequent anomaly associated with polydactyly.² To our knowledge, a case of polydactyly associated with hemivertebra has never been reported to date.

Hemivertebra, a type of vertebral segmentation anomaly, develops as the result of a defect in the formation of one half of a vertebral corpus. Rapid advances in genetic technology in recent years have shown that genetic factors may be involved in the development of congenital vertebral anomalies including hemivertebra. However, the complex relationship between this group of diseases and genetic etiologies has not yet been deciphered.³ Hemivertebra has been reported to occur in approximately 5 per 10 000 live births and constitutes a common cause of congenital scoliosis.⁴ On the other hand, a recent study showed that hemivertebra is present in about 0.27% of asymptomatic individuals.⁵ In more than 70% of cases, other congenital anomalies, especially other spinal anomalies, accompany hemivertebra. Dysplastic radii and cleft lip and plate are the other musculoskeletal anomalies reported to accompany the disorder.⁶ We reported the first case of polydactyly in association with hemivertebra. Our case shows that rigorous prenatal ultrasonographic evaluation and detailed neonatal physical examination are necessary to detect possible additional abnormalities in cases with a recognisable skeletal anomaly.

Figure 1 Anteroposterior (A) and lateral (B) radiographs of the right hand demonstrate duplication of both the proximal and distal phalanges of the thumb, consistent with Wassel type IV polydactyly.

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Learning points

- ▶ Polydactyly occurs as an isolated anomaly in the majority of cases.
- ▶ Hemivertebra is accompanied by other congenital anomalies, especially other spinal anomalies, in more than 70% of cases.
- ▶ Hemivertebra may accompany polydactyly.

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