Case report

Rhombencephalosynapsis associated with hand anomalies

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Abstract. A case of rhombencephalosynapsis, a very rare disorder characterized by agenesis or hypogenesis of the cerebellar vermis and fusion of the cerebellar hemispheres, is reported with magnetic resonance imaging features. Radiographs showed anomalies in both hands; namely phalangeal hypoplasia and occult polydactyly in the right hand and syndactyly in the left, previously unreported in association with this disorder.

Rhombencephalosynapsis is a very rare congenital malformation of the posterior fossa consisting of vermis agenesis or severe hypogenesis, fusion of the cerebellar hemispheres and apposition or fusion of the dentate nuclei [1]. To date, 24 cases have been reported, including the original description by Obersteiner in 1914 [2–5]. There is also a report of antenatal diagnosis of rhombencephalosynapsis at 18 weeks of gestation by ultrasound examination, prior to termination of pregnancy [6]. Associated anomalies within and outside the central nervous system have been reported in some cases with this disorder [1, 3, 7]. We present a case of rhombencephalosynapsis with phalangeal hypoplasia and occult polydactyly in one hand and syndactyly in the other, both conditions being reported for the first time in association with this anomaly.

Case report

A 17-month-old girl presented with irritability and developmental delay. She was the product of a full term pregnancy and spontaneous vaginal delivery, with a birth weight of 3800 g. The prenatal course was uneventful. This was the second pregnancy for her 28-year-old mother; the first child, a 4.5-year-old boy, was normal. There was no parental consanguinity. She was unable to sit unsupported at 10 months. She could step on her toes but was unable to walk heel-to-toe. Cerebellar tests were normal and there was no ataxia, tremor or nystagmus. She had low set ears. Her right hand was smaller than normal and there was syndactyly of the fourth and fifth fingers of her left hand. The right foot was mildly everted.

Cranial magnetic resonance imaging showed agenesis of the cerebellar vermis with fusion of the cerebellar hemispheres (Figure 1). Convergence of the dentate nuclei formed a horseshoe shaped arc across the midline posterior to the fourth ventricle, giving a “keyhole” shape to it. On radiographs phalanges of all fingers of the right hand (both phalanges of the thumb and especially the middle and distal phalanges of the other fingers) were smaller than normal and there was a sixth hypo-plastic metacarpal bone on the hypothenar side of this hand (Figure 2a). There was syndactyly of the fourth and fifth fingers of the left hand, no osseous fusion being present (Figure 2b).

Discussion

Rhombencephalosynapsis is characterized by agenesis or severe hypogenesis of the cerebellar vermis and apposition or fusion of the dentate nuclei, resulting in the appearance of a single-lobed cerebellum and “keyhole” shaped fourth ventricle on imaging [8]. Although it is extremely rare, rhombencephalosynapsis is being recognized with increasing frequency following the advent of MRI. This is probably due to the excellent morphological detail of posterior fossa structures provided by MRI.

Outside the central nervous system, the musculoskeletal system is by far the commonest site of associated anomalies in cases with rhombencephalosynapsis [1, 3, 7]. In one case, an infant girl who died shortly after birth due to severe respiratory distress and bradycardia, there was hypoplasia of the left radius, absence of the left thumb and first metacarpal bone of the right hand and multiple segmentation defects of the cervical and thoracic vertebrae and left upper ribs [7], as well as cardiovascular, respiratory and urinary anomalies in association with rhombencephalosynapsis.
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Figure 1. T1 weighted (a) and T2 weighted (b) transverse MR images of the posterior fossa show cerebellar hemispheric fusion with agenesis of the cerebellar vermis. Note the “keyhole” appearance of the fourth ventricle on (b).

Figure 2. Anteroposterior radiographs of the right (a) and left (b) hands show phalangeal hypoplasia in all fingers of the right hand with a hypoplastic sixth metacarpal bone (a, arrow) and syndactyly of the fourth and fifth fingers in the left hand.

Another patient, a 12-year-old boy, had a high arched palate, mild metatarsus varus deformity and clinodactyly of the fifth fingers in association with rhombencephalosynapsis [1]. High arched palate, which was not present in this case, has been reported in two other patients [1, 3]. Low set ears, a feature of the present case, have also been reported in two other patients with rhombencephalosynapsis, one of whom also had a deformed occipital bone [1].

Phalangeal hypoplasia, polydactyly and syndactyly have not been previously reported in association with rhombencephalosynapsis. Embryologically, some sort of an insult between the fifth and eighth weeks of gestation seems to result in the absence or hypoplasia of the cerebellar vermis. This, in turn, permits the primordia for the cerebellar hemispheres to fuse together [8]. Phalanges, on the other hand, are formed near the end of the second month of gestation and
syndactyly results from the failure of the breakdown of mesenchyme between the prospective digits in the hand plate between the sixth and eighth weeks of gestation [9]. It is therefore likely that a disturbance of organogenesis sometime during the second half of the second month of gestation is responsible for the anomalies in the present case.

In conclusion, although there are few reported cases of rhombencephalosynapsis, musculoskeletal anomalies seem to be a relatively common association and a radiological survey of the skeletal system should be made in affected cases.

References