CASE REPORT/KAZUISTIKA

Thoracomelia in Poland Anomaly

Thorakomelie u Polandova syndromu

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SUMMARY

Poland Anomaly (PA) represents pectoral muscle hypoplasia in combination with various forms of hand anomalies. We report a case of PA with pectoral hypoplasia, an upper limb deficiency and an accessory extremity/digit on the ipsilateral thoracic wall (thoracomelia). To our knowledge, this is the first reported case of thoracomelia in PA.

Key words: ulnar aplasia, thoracomelia, Poland anomaly.

INTRODUCTION

Alfred Poland described Poland Anomaly (PA) as a medical student in 1841, reporting a case of pectoral muscle agenesis in combination with other muscle deficiencies and ipsilateral brachysyndactyly of the hand (13). The incidence has been reported between 1/30,000 and 1/32,000 (6, 12). Poland syndrome usually presents in an isolated form, although associations to other syndromes exist, such as Moebius, Kippel-Feil, Pierre-Robin syndrome (5). The exact mechanisms underlying PA remain unclear, but a hypothesis of vascular insult during early embryonic development has been proposed (5). Initially, a limb bud has a capillary network, the arterial system appears afterwards. The median artery is the principal blood supply to the hand, but regresses as the radial and ulnar arteries take over, and the median artery end supplying blood to the median nerve (5). A mild ischemic insult disrupting the development of the sternocostal head of the pectoralis major muscle, finger separation and chondrification of the middle phalanges has been proposed (1). Familial recurrence has been noted in 10% of cases, displaying inheritance patterns like autosomal dominant with incomplete penetrance, autosomal recessive as well as X-linked (4). Thus, the risk for a person with PA to have children with PA is increased compared to the general population. PA is more common in males compared to females (ratio 1.7:1) and the right side is more often affected than the left (ratio 1.6:1)(14). Rib agenesis and/or rib hypoplasia has been noted in 25%. In addition, pectus excavatum/carinatum, spine deviations and renal malformations have been described in some cases (14). The diagnosis is established by physical evaluation (1). In the revised Oberg-Manske-Tonkin (OMT) classification of congenital upper limb anomalies (CULA), PA is placed in group IA, 1iia abnormal axis formation, proximo-distal axis, symbrachydactyly spectrum with ectodermal elements (7). Subclassifications of PA have been proposed by Al-Quattan 2001 in seven subtypes, and by Romanini et al. in 2018 (three subtypes)(1, 14).

Polymelia is a rare CULA where a supernumerary limb is present. If attached to the thoracic wall, the term *thoracomelia* can be used. Case reports on this condition are scarce. One recent article reports a case of lower limb polymelia in a female child in India, another reports a case of thoracomelia in a 1.5-year-old Nepalese child (9, 17).

To our knowledge, thoracomelia has not previously been reported in association with PA. Here we present a case of PA in combination with thoracomelia.

CASE PRESENTATION

A baby girl was born as a first child at 39 + 0 gestational weeks with a Caesarean section due to the fetus being in a breech position. Apgar score 9-10-10 (at 1, 5 and 10 minutes post-partum), birth weight 3230 grams (-0.5 SD), length 50 cm

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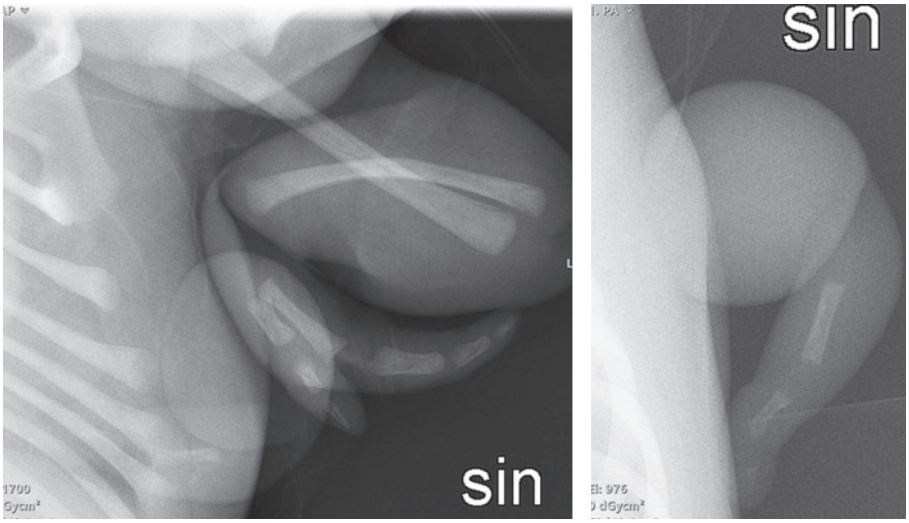

Fig. 1. **Radiographs at age 1 week with ulnar ray deficiency and accessory digit/limb on thoracic wall.**

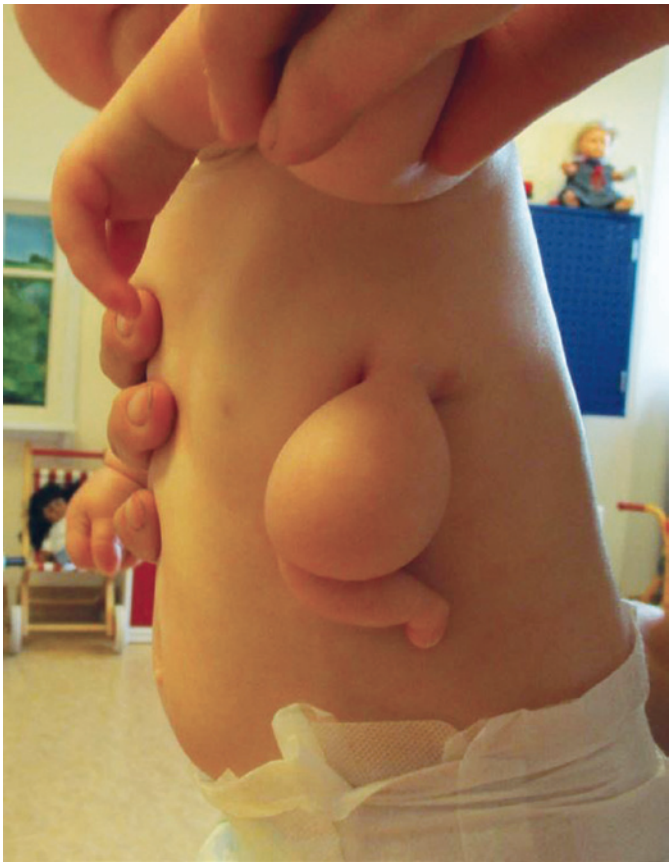

Fig. 2. **Photo of accessory digit/limb at age 4.5 months.**

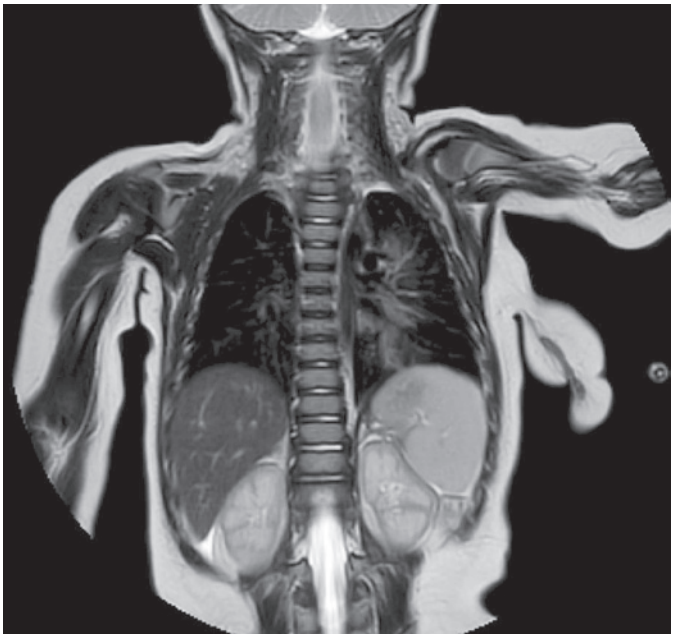

Fig. 3. **MRI of thoracic wall with attached digit/limb.**

(50th percentile), Occipital Frontal Circumference (OFC) was 35 cm (50th percentile), and no family history of hand anomalies or exposure to potential teratogens. No prenatal studies had been performed. She presented with a left side upper limb deficiency with no ulna and the three ulnar rays of the hand missing, pectoral muscle hypoplasia and a small limb/accessory digit attached to the thoracic wall (Fig. 1 and 2). The left and right nipple/areola complex were similar. The combination of findings is somewhat atypical, but probably fits the clinical diagnosis of PA best. The radiographs demonstrated the presence of three phalanges in the accessory digit. A nail was present distally. As the elbow was in flexion, gentle stretching of the elbow by the parents was commenced in combination with a semicircular night splint for the elbow. At age 9 months, she underwent a thorax MRI which showed hypoplasia of the left thorax but all ribs were present. There was also severe hypoplasia of the pectoral muscles and the glenoid of the humerus (Fig. 3). Due to the small size of the child, the different parts of the pectoral musculature were difficult to identify

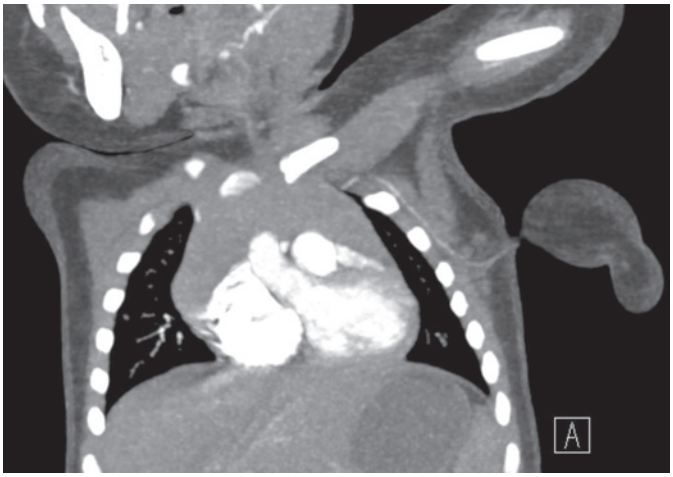

Fig. 4. **CT angiography showing that the accessory digit/limb is supplied by the internal mammary artery.**

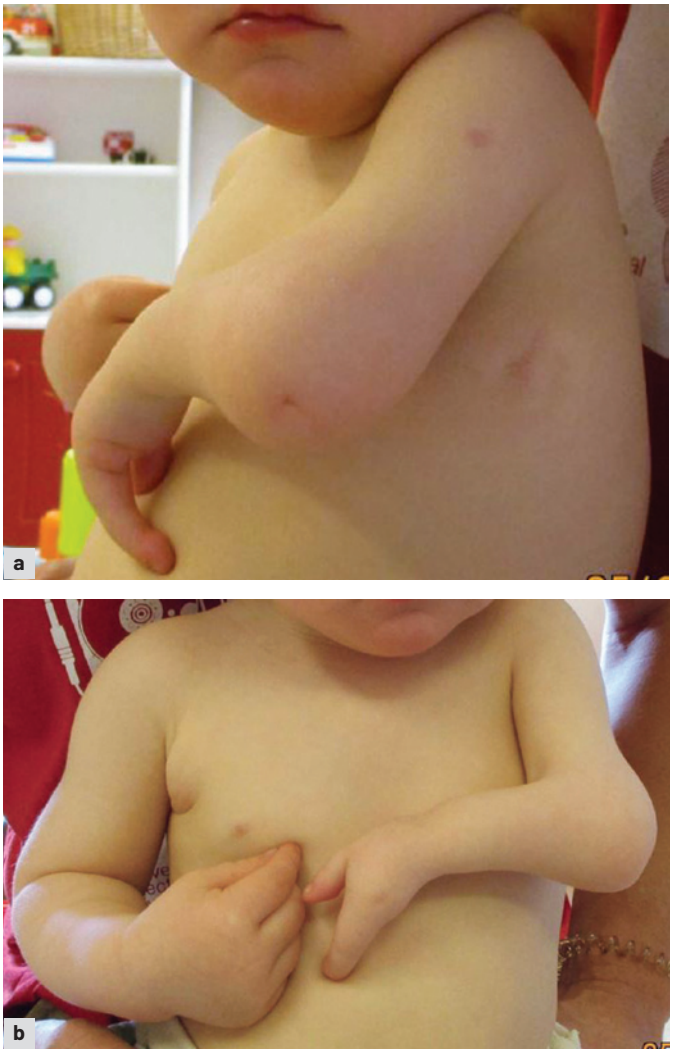

Fig. 5. **Photographs at age 16 months after removal of accessory digit/limb.**

separately for the radiologist. The blood vessels supplying the accessory digit were difficult to visualize. Therefore, a CT angiography was done at age 10.5 months. This showed that the vascular supply to the accessory digit came from the internal mammary artery, which in turn had a normal connection to the left subclavian artery (Fig. 4). She underwent surgery at age 11.5 months where the accessory digit was surgically removed without complications. A follow-up at age 16 months showed good healing and a flexed resting position of the elbow (Fig. 5). She has continued with a night splint for the elbow and at the latest follow-up (age 5 years) had a grip between the thumb and index of the hypoplastic left hand and is able to grasp and hold small objects. Radiographs at age 5 show mineralization of the humeral head which is correctly placed in the glenohumeral articulation and a complete absence of the ulna (Fig. 6). The elbow has full active flexion and a passive extension deficit of 90°, likely related to the absent ulna and triceps insertion. Night splinting of the elbow is likely to continue until she is fully grown. Based on normal developmental milestones up to 5 years of age, physical exams and the CT and MRI thorax scans, the child was assessed as otherwise well-developed physically and mentally with no other associated malformations.

DISCUSSION

To our knowledge, this is the first case of thoracomelia reported in PA. According to the Al-Qattan classification, this patient most likely has PA type 4E, but type 7 (phocomelia-like deficiency)

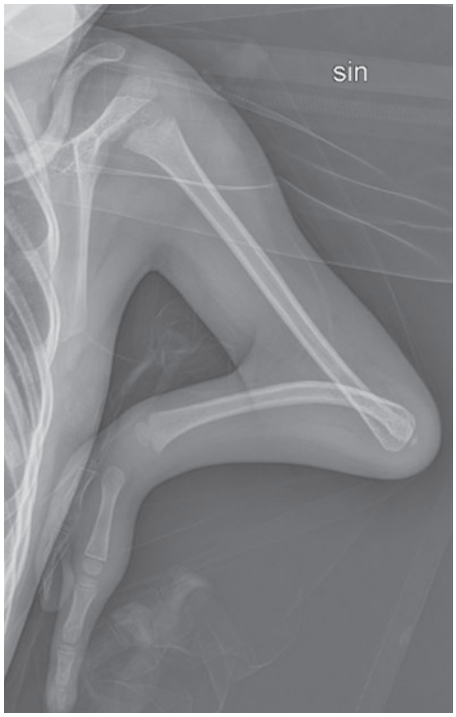

Fig. 6. **Radiograph at age 5 years showing mineralization of the humeral head and absence of the ulna.**

could also be considered (2). As pointed out by Goldfarb et al., upper extremity phocomelia could represent a spectrum of severe longitudinal dysplasia (8). The PA-classification proposed by Romanini et al. indicates that our patient has a type 3 PS (complete form) (14). As for the updated OMT classification, this patients´clinical findings indicate PS IA, 1iia, the arm probably best corresponds to IA 2ii (7). It should be noted that Alfred Poland´s initial description was of pectoral muscle deficiency ad ipsilateral symbrachydactyly. In addition, the use of the diagnosis PS is widely variable, and up to 10 different definitions have been described. Pectoral muscle hypoplasia is not distinctive for Poland syndrome alone but is also present in syndromes with other associated anomalies with a recognized genetic cause (3). Polymelia or thoracomelia in itself is extremely rare and to our knowledge has been reported only in a few case reports (9, 10, 17). The upper limb develops during the fourth week of gestation by activation of mesenchymal cells in the lateral plate mesoderms somatopleuric layer. At the apex of the limb bud the thickened ectoderm forms the apical ectodermal ridge (AER), which is induced by by parachrine factors from the mesenchyme. FGF-8 initiates proximodistal axis growth and differentiation. The TBX5 gene belongs to a family of transcription factors known as T-box genes, believed to play a role in the regulation of embryonic development and also associated with thoracomelia. According to the Nosology of genetic skeletal disorders: 2023 revision, PA is classified in the 38th Limb hypoplasia-reduction defects group as NOS 38-0360 (15). A mutation in the WNT7A gene has been reported in limb reduction syndromes and mutations in genes such as WNT3A, FGF10, and SHH have been associated with limb malformations, including thoracomelia, in animal models and human studies (11). Genetic testing has not been undertaken in this patient, although whole exome sequencing has been suggested to the parents who are considering this option (16). At present, there is no support to conduct standard karyotype or array-CGH genetic test in routine clinical investigations (14), but this could be an option in this case as well as whole exome sequencing. In our opinion, a multidisciplinary approach is important to achieve optimal function and to provide support to the family in cases like these. ■

Ethical approval: The study was conducted in accordance with the Declaration of Helsinki (as revised in 2013). Oral and written informed consent has been given by the patient's parents. Radiographs and photos are anonymized.

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