

Supernumerary nipples, congenital scoliosis, spina bifida occulta, tethered cord and diastematomyelia

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Abstract. A 3-months old girl who presented to our clinic with accessory breast and had signs of supernumerary nipples, congenital scoliosis and diastematomyelia was presented as she didn't correspond to any known syndrome.

Key words: Supernumerary nipples, congenital scoliosis, diastematomyelia

1. Introduction

The frequency of isolated supernumerary areolae involving a nipple is 1.4% in the general population. There is a rare autosomal dominant form of isolated polythelia (1-3). In addition, it has been shown that supernumerary nipples are also associated with several syndromes including a lethal type of popliteal pterygium syndrome (4), the Simpson-Golabi-Behmel syndrome (5) and the Char syndrome (6). Recently, it has been reported that supernumerary nipples associated with Becker nevus syndrome also exist (7). In the literature, there are reports suggesting supernumerary nipples associated with spina bifida (8) and scoliosis (9).

2. Case report

A 3-months old girl presented to our clinic with accessory breast. The body weight, height and head circumference of the patient were 4900 g (10th percentile), 57 cm (10th-25th percentile) and 38 cm (3rd-10th percentile), respectively. There was no consanguinity between parents. The father was 27 years old, whereas the mother was 25 years old. The patient was the first child of the family. There was no pathological event during

prenatal, natal or postnatal period. On the physical examination, it was observed that there was round face, wide nose, micrognathia, prominent filtrum, small nose, supernumerary nipples on the left side, normal localization of right nipple, scoliosis, short neck and hemangioma at lumbosacral region (Figure 1, 2). No extremity anomaly or polydactyl were detected. Urogenital and rectal examinations were also normal. Complete blood count, liver and renal function tests were found to be normal. A 46, XX karyotype was detected. Scoliosis, rib agenesis and deformation (deformation at the ribs 3, 4 and 6, and agenesis at ribs 5 and 7) were observed on the whole-body bone X-ray evaluation (Figure 3). Abdominal and urinary sonographies were found to be normal. Cardiac echocardiography revealed normal findings. On MR imaging, lumbar spina bifida, tethered cord and diastematomyelia were detected (Figure 4). Cranial MR imaging was also normal.

3. Discussion

Supernumerary nipples aren't only seen as an isolated entity but also it can be familial or a component of a syndrome (1-9). Several syndromes have been defined in the literature, which are associated with supernumerary nipples (4-7). In our case, autosomal dominant form was excluded as none of the parents have supernumerary nipples. It is evident that it wasn't an isolated entity as associated anomalies were present. Supernumerary nipples are rare entities in general population and additional dysmorphic and development defects were present in this case; thus, we reviewed previously reported syndromes, which can be associated.

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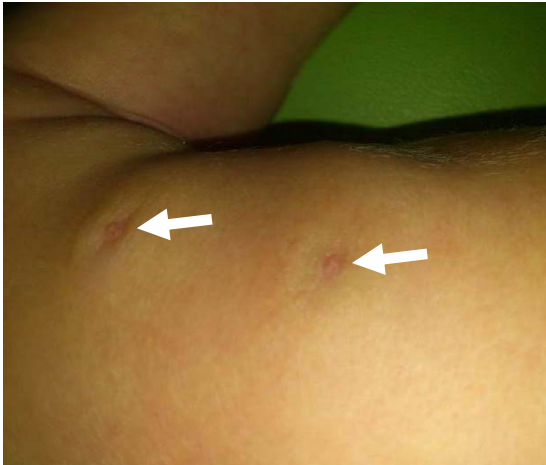


Fig. 1. Supernumerary nipples on the left side (arrows).



Fig. 2. Hemangioma at lumbosacral region (arrow).



Fig. 3. Scoliosis, rib agenesis and deformation in X ray.



Fig. 4. On MR imaging, lumbar spina bifida, tethered cord and diastometamyelia were detected.

Supernumerary nipples were reported in an Iranian case exhibiting features of Barsocas-Papas syndrome (4), which is characterized by multiple popliteal pterygia, ankyloblepharon, filiform bands between the jaws, cleft lip and palate, and syndactyly (10). The Barsocas-Papas syndrome (lethal popliteal pterygium syndrome) has autosomal recessive inheritance. However, our case had none of these features. Supernumerary nipples were also reported in a girl and her father with Char syndrome published in 2000. Our case had none of the main clinical

characteristics of Char syndrome such as typical face appearance, strabismus or foot anomalies. When our case was compared to the case Simpson-Golabi-Behmel syndrome associated with postaxial polydactyl, lack of congenital heart defect, crude facial appearance, pre- and post-natal overgrowth excluded this syndrome in our case (11).

It has been also reported that supernumerary nipples were associated to Becker Nevus Syndrome (7). This syndrome is an association of Becker's nevus with breast hypoplasia and other

anomalies; the most frequently seen anomalies are lumbar spina bifida, thoracic scoliosis, and pectus carinatum always at ipsilateral side with lesion (12). More recently, association of congenital scoliosis, supernumerary nipples and spina bifida occulta has been reported in 3 cases from India (13). In those cases, supernumerary nipples were at the ipsilateral side with rib anomalies. However, in our case, there was tethered cord and diastematomyelia as well as these findings. The association of congenital supernumerary nipples, spina bifida and scoliosis in addition to development of Becker nevus at adolescent age has been reported in the Becker nevus syndrome (7). However, the Becker nevus syndrome doesn't include typical facial appearance. The facial appearance was common in our case and those reported from India. In our case, there were supernumerary nipples, congenital scoliosis, spina bifida, tethered cord and diastematomyelia as well as round face, wide nose, micrognathia and hemangioma at lumbosacral region, while filtrum was prominent.

It was thought that the present case, which didn't correspond to previously known syndromes, could be a distinct syndrome as it exhibited similar features to those reported from India and was discussed here as it is an interesting entity. We think that the component of this syndrome can be listed as supernumerary nipples, ipsilateral rib defects, scoliosis and medulla spinalis anomalies, and that syndrome can be termed as "Supernumerary nipples, ipsilateral costa deformity, scoliosis and congenital medulla spinalis malformations".

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