Trisomy 18 syndrome, also called Edwards syndrome, was first described in 1960 as showing trisomy of chromosome 18 in a newborn with multiple malformations. Trisomy 18 syndrome occurs in newborns at a rate of about 1/5000 and about 80% of newborns affected by this disorder are female. This chromosomal condition is associated with severe intellectual disability and multiple physical abnormalities. More than 130 anomalies are described in cases of trisomy 18 syndrome. Smith and Schinzel revisited the trisomy 18 syndrome and reported the most frequently seen extremity anomalies include clenched hand with overlapping digits, nail hypoplasia, dorsiﬁlexed short hallux, ulnar, radial deviation of hand, hypoplasic or absent thumb, equinovarus, rocker bottom feet and syndactyly of the second and third toes.1,2 There are only 5 cases of ectrodactyly in the literature. We present a trisomy 18 syndrome case with unilateral ectrodactyly of the left foot, which is rarely seen.

CASE REPORT

A male infant was born to a 28-year mother by elective caesarean section with an APGAR score three at the first minute and five at the fifth minute. After a difﬁcult resuscitation in the delivery room, he was transferred to our newborn intensive care unit owing to breathing difﬁculty and multiple congenital anomalies. Endotracheal intubation was not required.

The infant birth weight was 1930 g (small for gestational age), head circumference 32 cm (10 - 25 (th) percentile), length 45 cm (< 10 (th) percentile). He presented low birthweight, a small, abnormally shaped head, low-set ears, hypoplasia of orbital ridges with epicanthal folds and narrow palpebral ﬁssures, a small jaw and mouth with high palate, micrognathia, bilateral syndactyly, aplasia and hypoplasia of the foot digits, unilateral ectrodactyly of the left foot and a prominently dorsiﬁxed hallux.

Figure 1a: Feet of the patient; syndactyly, aplasia and hypoplasia of the digits and ectrodactyly of the left foot.

Figure 1b: X-ray of the cleft foot; showing fusion of the ﬁrst, second digits and third, fourth digits. The absence of the second and fourth phalanges can also be seen.

Figure 2: The cytogenetic analysis of the patient.
hallux, clenched hand with overlapping fingers and general hypertonia (Figure 1a and b). Echocardiography showed patent ductus arteriosus and mild pulmonary stenosis. He required continuous tube feeding from the first few days of life.

Chromosome analysis of a peripheral blood sample showed 47, XX+18 (Trisomy 18) karyotype (Figure 2).

DISCUSSION

Ectrodactyly, also known as split-hand/split-foot malformation (SHFM), can occur as an isolated entity or as part of a syndrome. Both forms are frequently found in association with chromosomal rearrangements and mutations that fail to maintain the median apical ectodermal ridge signal. 3 Ectrodactyly is characterized by underdeveloped or absent central digital rays, clefts of hands and feet, and variable syndactyly of the remaining digits.

Extremity anomalies are frequent in cases with trisomy 18 syndrome and Smith’s review categorizes them according to frequency.2 Clenched hand, tendency for overlap of index finger over fourth finger, absence of distal crease on fifth finger with or without distal crease on third and fourth fingers, hypoplasia of nails and short hallux; also, ulnar and radial deviation of the hand, hypoplastic to absent thumb, simian crease, equinovarus and rocker bottom feet are found in 10 - 50% of cases. Smith and Schinzel conclude that ectrodactyly is seen in < 10% of cases.1,2 This is an interesting comment because there are only 5 cases in the literature and the authors had not found any case with ectrodactyly in their unpublished 96 cases of trisomy 18 syndrome. Rosa et al. reported that only one patient with ectrodactyly was found in 50 cases of trisomy 18 syndrome.4 There are trisomy 18 syndrome cases with severe cleft foot anomalies in the literature.5 Butler et al. and Moerman reported the first case of trisomy 18 syndrome with cleft foot;6,7 there are three other reported cases.8-10 The present case is very rare form of trisomy 18 syndrome associated with ectrodactyly.

REFERENCES