What is your diagnosis?

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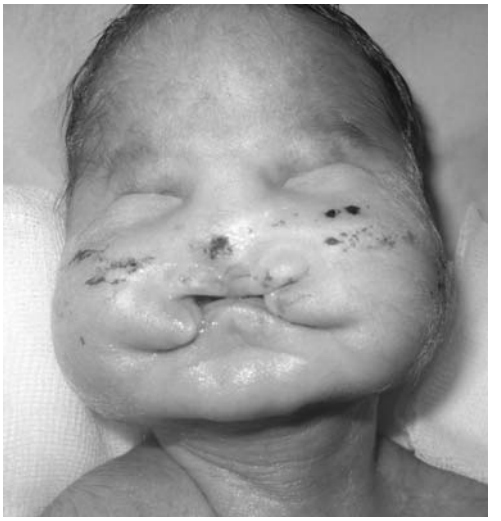
A male infant, weighing 1080 g was delivered at 26 gestational weeks. He had a congenital nasal aplasia. He was transferred to neonatal intensive care unit with the diagnosis of respiratory distress syndrome. He was immediately intubated and surfactant was given by rapid bolus instillation. The initial clinical examination revealed facial dysmorphism including a prominent wide forehead, bushy eyebrows, wide anterior fontanel, small palpebral fissures, ocular hypertelorism, high arched palate, nasal aplasia (Figure 1), low-set ears, micrognathia, bilateral clenched hands with over lapping fingers, nail hypoplasia, rocker-bottom feet and penile hypospadius. He was weaned progressively from mechanical ventilation and was extubated after 18 hours. But he passed away because of major congenital cardiac defect at 7 days old.

Figure 1: Nasal aplasia.



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Diagnosis: Edwards syndrome (Trisomy 18)

Trisomy 18 syndrome is the second most common chromosomal aberration syndrome caused by an extra number 18 chromosome. It was first described in 1960 by Edwards et al.; therefore, it is also named as Edward's syndrome (1) . Its incidence is given as 0.3 per 1,000 live births (2). A variety of anatomic abnormalities involving almost all organ systems have been noted. Our patient has a prominent wide forehead, bushy eyebrows, wide anterior fontanel, small palpebral fissures, ocular hypertelorism, high arched palate, nasal aplasia, low-set ears, micrognathia, bilateral clenched hands with over lapping fingers, nail hypoplasia, rocker-bottom feet and penile hypospadius. Nasal aplasia has been reported very rare in Edward syndrome. Our patient's chromosome analysis of the peripheral blood confirmed trisomy of chromosome 18 (47, XY+18).

REFERENCES

1. Edwards JH, Harnden DG, Cameron AH, Crosse VM, Wolff OH: A new trisomic syndrome. Lancet, 1: 787-790,1960

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