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# Craniofacial Microsomia: An Unusual Finding in Newborn

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## Case

A late preterm male baby was born normal vaginal delivery to G2 A1 mother. The infant had normal Apgar scores of 7/8/8 at 1, 5 and 10 minute respectively. At birth baby was diagnosed to have preauricular tags (Figure 1), left mandibular hypoplasia (Figures 2 and 3), hypertelorism, high arched palate. There was no genital, crease or anorectal abnormality. The infant was evaluated with whole body X-ray, USG-KUB, head ultrasound and echocardiography. Echocardiography showed normal study and ultrasound KUB was suggestive of prominent left pelvicalyceal system with no other significant abnormality. Head ultrasound done was not suggestive of nay abnormality. X-ray of left mandibular taken was suggestive of congenital agenesis of left mandibular bone (Figure 4). The provisional diagnosis of craniofacial microsomia was made.

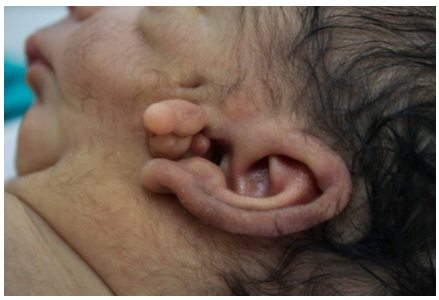

**Figure 1:** Left preauricular tag in the neonate

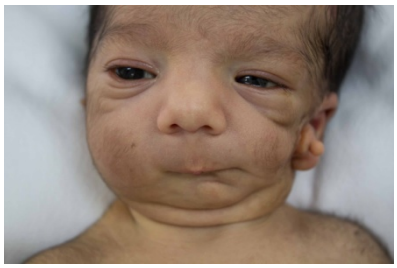

**Figure 2:** Hypoplasia of the left mandibular with deviation of mouth to left because of mandibular hypoplasia (Note: The associated preauricular tag)

**Figure 3:** Hypoplasia of the left mandibular with deviation of mouth to left because of mandibular hypoplasia (Note: The associated preauricular tag)

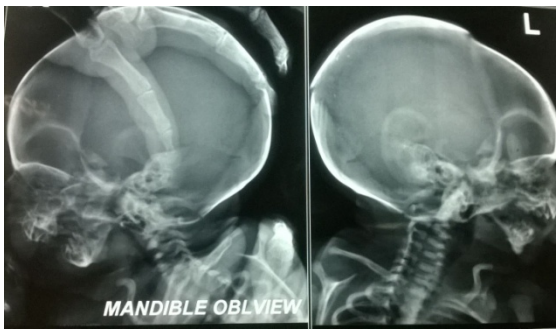


**Figure 4:** Lateral X-ray of left mandibular taken was suggestive of congenital agenesis of left mandibular bone

## Discussion

Craniofacial microsomia (CFM) includes a variety of malformations primarily due to embryologically derived defect in development of the first and second branchial arches. It is also known as hemifacial microsomia as there is underdevelopment of one side of branchial arches. The causes is unknown with no specific pattern of inheritance [1,2]. The cause is thought to be abnormal changes in blood vessels that lead to disruption of the blood supply during fetal development at around 30-45 days of gestation which leads to underdevelopment of the structures derived from the first and second branchial arches [3].

The clinical features includes facial asymmetry because of maxillary and/or mandibular hypoplasia, usually right sided hypoplasia;

preauricular or facial tags; ear malformations such as microtia, anotia, or aural atresia; frontal bossing; diminished or absent parotid secretion, tongue anomaly in form of malfunction of taste sensation and hearing loss. It has variety of presentation which can range from subtle facial asymmetry with a small skin tag in front of anormal ear to rare bilateral involvement, microtia/anotia with atresia of the ear canals, microphthalmia, and respiratory compromise from severe mandibular hypoplasia. The other associated malformations includes IUGR, small for gestation age, hemi vertebrae or agenesis of vertebrae, usually cervical (40-60%), ophthalmological including epibulbar dermoid and/or lipodermoid, strabismus, coloboma of the superior lid, microphthalmia, cleft lip, cleft palate. The cardiac malformation includes ventricular septal defect; patent ductus arteriosus; tetralogy of Fallot; coarctation of the descending aorta. The associated pulmonary malformations can be pulmonary agenesis or hypoplasia of the lung whereas renal includes ectopic and/or fused kidneys, renal agenesis, double ureter. Nervous system associated malformations includes mental retardation; occipital encephalocele; hydrocephalus; agenesis of corpus callosum [4,5]. Differential diagnosis includes [6]:

- Treacher-Collins Syndrome
- Oculo-auriculo-vertebral spectrum
- Otomandibulardysostosis
- Facio-auriculo-vertebral syndrome
- Lateral facial dysplasia
- CHARGE Association
- VACTERL Association
- Townes-Brocks Syndrome
- Branchio-Oto-Renal (BOR) Syndrome
- Chromosomal disorders (including Trisomy 9 Mosaicism, Trisomy 18, Ring 21 Chromosome, and Trisomy 22)
- Teratogen exposure

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