

Cerebrocostomandibular Syndrome

*Dr Anupriya Gupta
North Middlesex University Hospital, London, U.K.*

Abstract

We report a postnatal diagnosis of cerebrocostomandibular Syndrome (CCMS) in a new born after a low risk pregnancy. The dating scan, combined test and anomaly scans were normal. Polyhydramnios was noted at 28 weeks (amniotic fluid index: 28.2mm). The mother delivered by an emergency Caesarean for failure to progress.

The female baby was born through thick meconium and required 3-4 mins of resuscitation. She was noted to have dysmorphic features (micrognathia, submucosal cleft palate, bilateral sacral dimples, left ear pit, anteriorly positioned anus). Further evaluation made a diagnosis of CCMS secondary to a de novo mutation. We discuss this rare syndrome and undertake a literature search for isolated micrognathia. The anomaly check in the UK follows the National screening committee and Fetal anomaly screening programme (FASP) guidelines. FASP recommends a mid-pregnancy scan which is undertaken between 18+0 to 20+6 weeks to screen for major fetal anomalies. The main structures to be assessed between these weeks are defined. Abnormalities of these structures can indicate a number of specific conditions. Coronal view of lips with nasal tip are viewed but not the sagittal view of the face. If micrognathia is the only sonographic finding identified, physicians and families should be prepared for possible respiratory difficulty at delivery, the presence of a cleft palate, and/or

Developmentaldelay



Biography

Dr Gupta has completed her MRCOG(U.K) and Masters in Obstetrics and Gynaecology from Devi Ahilya Viswavidhyalaya. She is a junior doctor at North Middlesex University Hospital, London, U.K. She has been actively publishing articles in peer reviewed journals.

| Presenting | author | details |
|------------|-------------------|-------------------------|
| Full name: | Anupriya Gupta | |
| Contact | number: | +44-7587480306 |
| Twitter | account: | - |
| Linked In | account: | anupriya.0016@gmail.com |
| Session | name/ | number: Track 5 |
| Category: | Oral presentation | |