

Aetiology

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Non-syndromic cleft lip and/or palate may present as new diagnosis within a family or with a clear family history. A family history of cleft lip and palate in which a first-degree relative is affected increases the risk of subsequent cleft cases in the family, supporting the theory that there are underlying genetic mutations contributing to the aetiology. Isolated cleft palate is more commonly associated with a syndrome than cleft lip and palate and isolated cleft lip. Over 150 named syndromes are associated with cleft lip and palate, although Stickler (ophthalmic and musculoskeletal abnormalities), DiGeorge (cardiac/thymic anomalies), Down, Apert and Treacher Collins syndromes are most frequently encountered. aetiology of cleft lip and/or palate, including maternal epilepsy (and associated medication) and drugs (e.g. steroids, diazepam, sodium valproate and phenytoin). The role of antenatal folic acid supplements in preventing cleft lip and/or palate remains equivocal. Pierre Robin sequence is a condition worth considering in specific terms. This sequence comprises isolated cleft palate, retrognathia and a posteriorly displaced tongue (glossoptosis), which is associated with early airway and feeding difficulties. Although airway obstruction does not commonly occur in babies with a non-syndromic cleft lip and/or palate, in babies with an airway obstruction, e.g. Pierre Robin sequence hypoxic episodes during sleep and feeding can be life-threatening. Intermittent airway obstruction is more frequent and is managed conservatively. In more severe cases the children will often require adjunctive support for their airway compromise such as supplemental oxygen, nasopharyngeal airway and even tracheostomy. More controversially, surgical adhesion of the tongue to the lower lip (labioglossopexy) in the first few days after birth is an alternative but less commonly practised method of management. Mandibular distraction surgery has advocates but numerous attempts at developing a consensus view in support of this procedure for airway compromise in cleft have been unsuccessful. Summary box 50.2 Aetiology of cleft lip and palate /uni25CF /uni25CF /uni25CF

The cause of cleft lip and/or palate is multifactorial Most cases occur without a clear family history or known risk factors Clefts can be associated with many craniofacial/medical syndromes

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