La Sindrome di Treacher Collins

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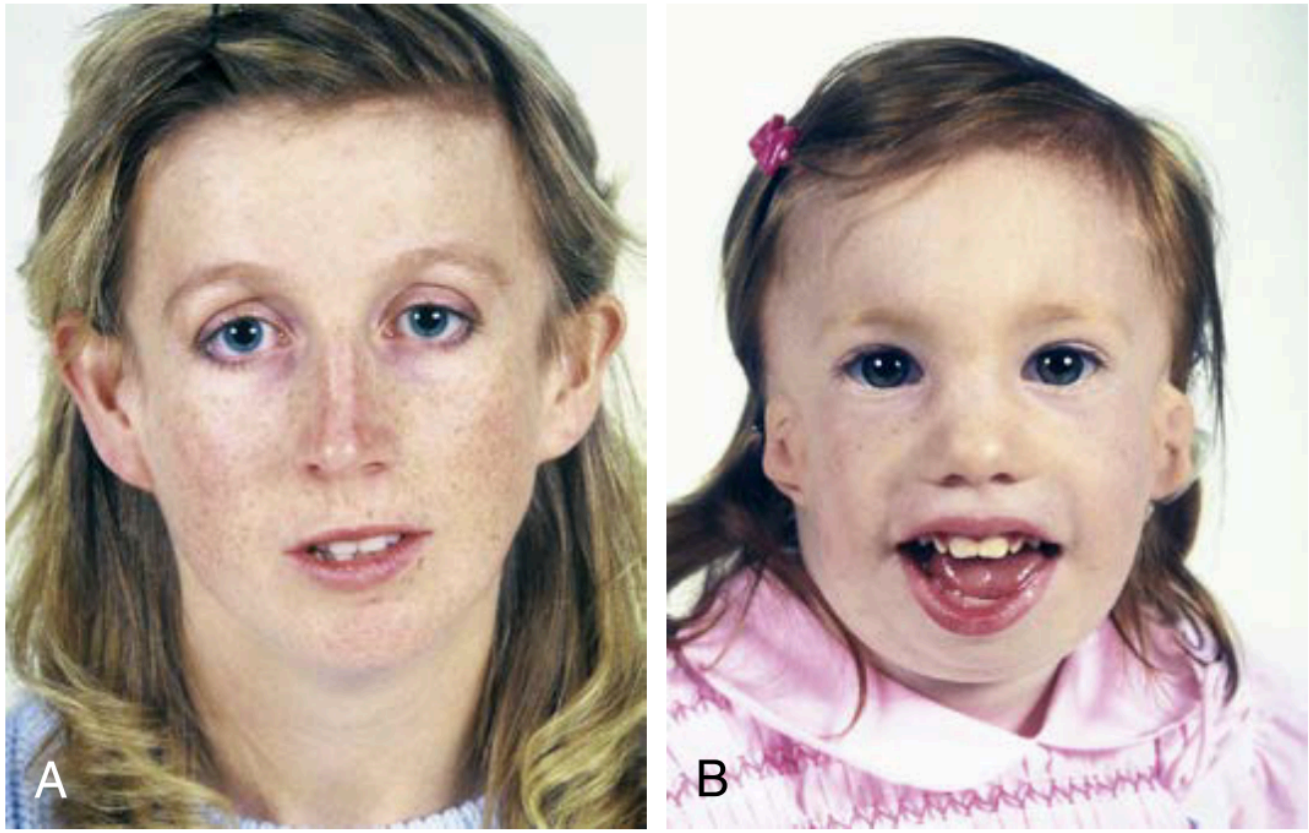
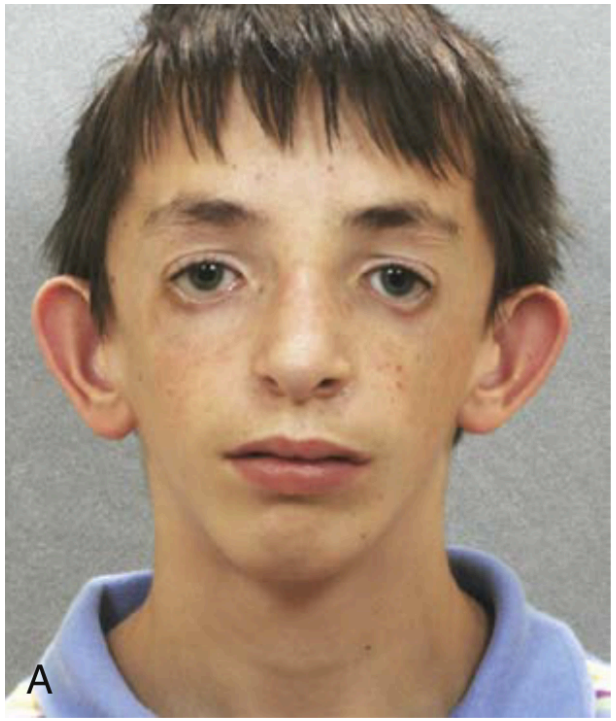
Sindrome di Treacher Collins

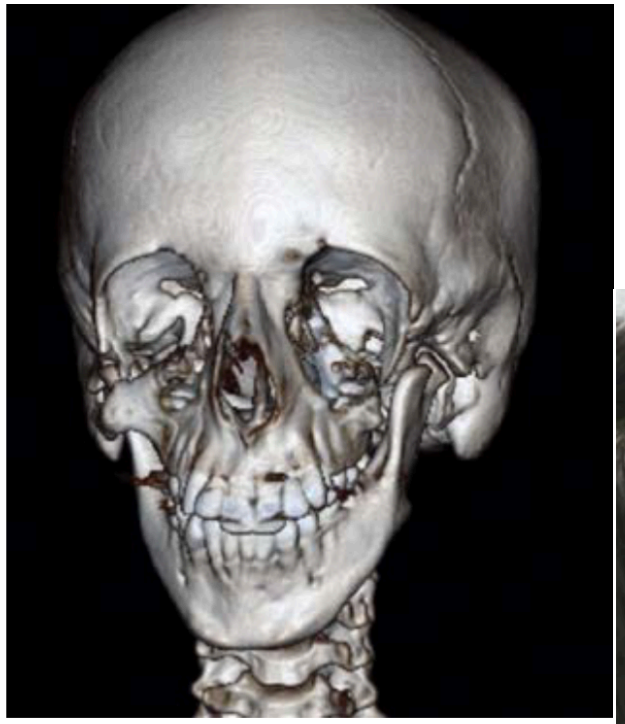
- Autosomico dominante a penetranza variabile
- Mutazione del gene TCOF1
- 1/25000-50000 nascite
- Ipoplasia o aplasia malare
- Convessità facciale
- Ipoplasia mandibolare
- Ipoplasia o aplasia condilare
- Mento retratto
- Plica antimongolica e coloboma
- Orecchie normali o deformate o assenti
- Atresia del canale uditivo-sordità
- Labiopalatoschisi

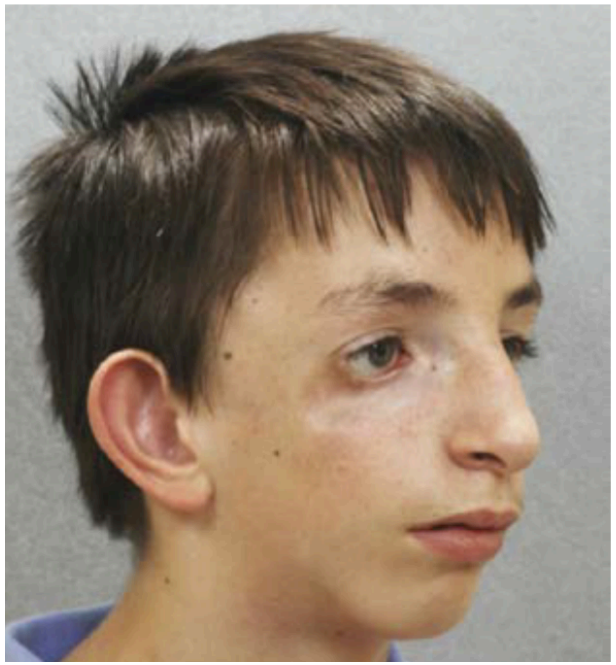
T.C.

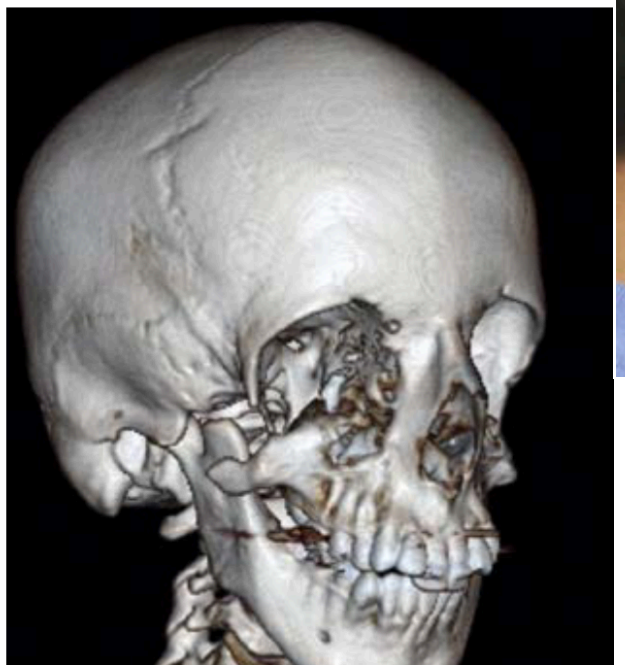


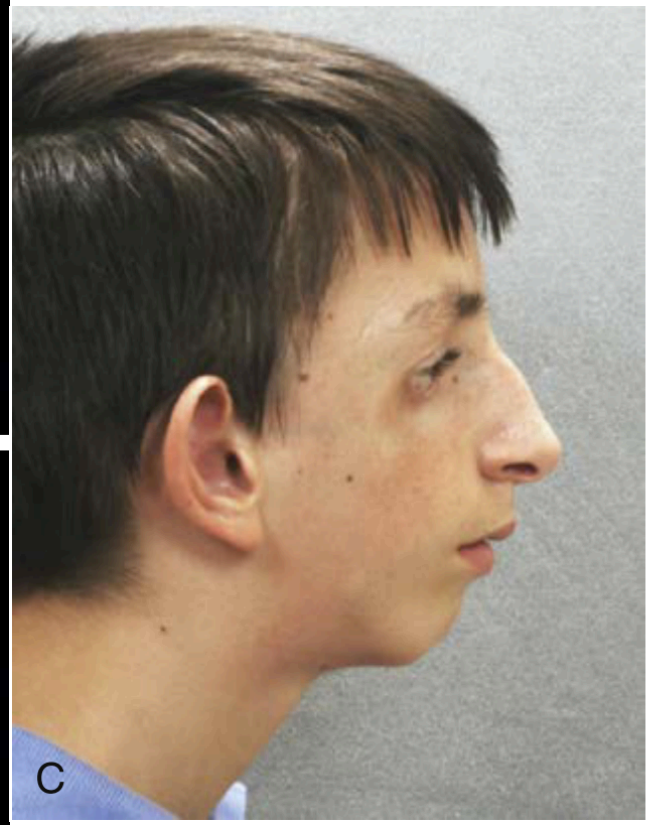
• **Figure 40-2** This mother **(A)** and daughter **(B)** demonstrate the extent of variation in the expression of Treacher Collins syndrome within a family. The mother was not aware that she carried the Treacher Collins gene until after the birth of her daughter. (From Posnick JC: Treacher Collins syndrome: perspectives in evaluation and treatment, *J Oral Maxillofac Surg* 55:1120, 1997.)

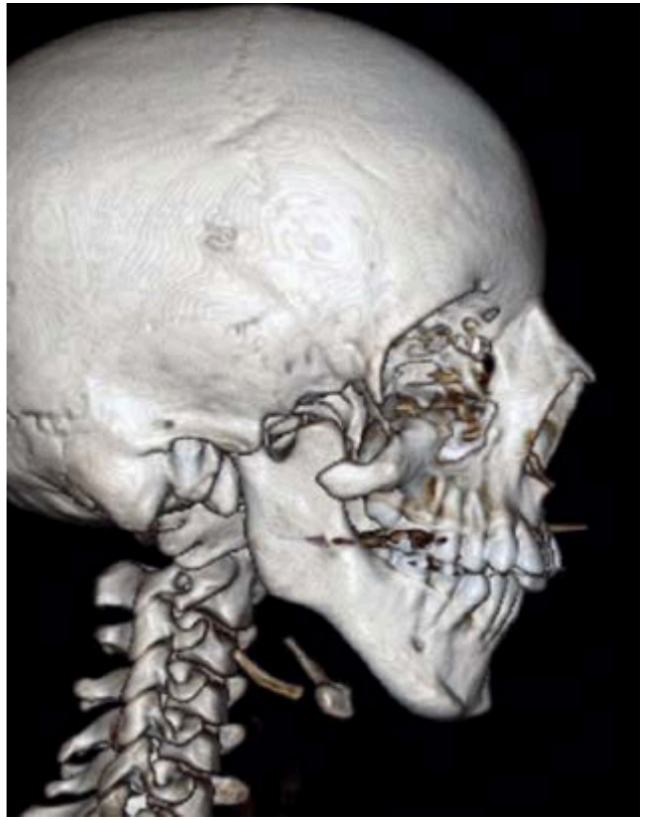

A





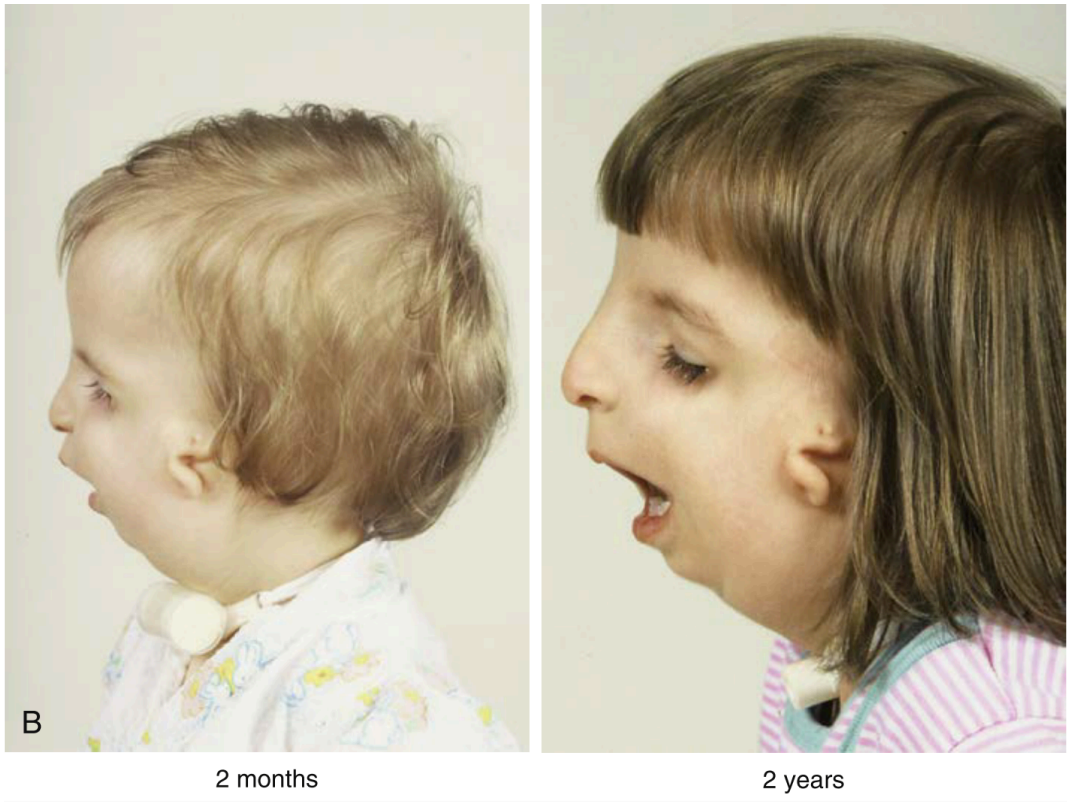



C



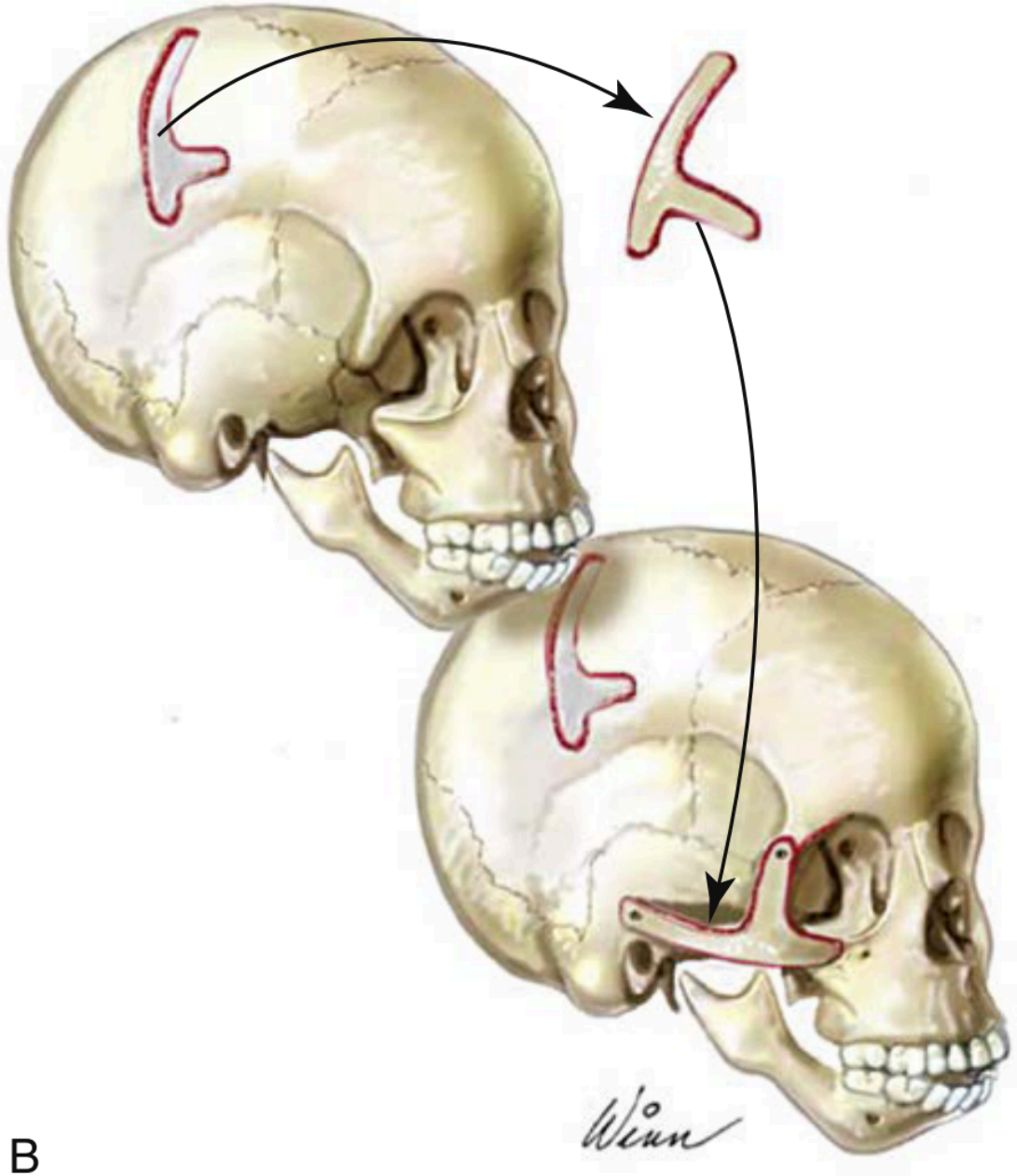
Sindrome di Treacher Collins

- Problemi alla nascita: apertura delle vie aeree compromesse dall'ipoplasia mascellare e malare con eventuale atresia delle coane, aggravate dall'ipoplasia mascellare e da tracheomalacia, deglutizione, udito, visione (ulcerazioni corneali), sostegno alla famiglia.
- Problemi successivi: TCS resta stabile senza aggravarsi con la crescita

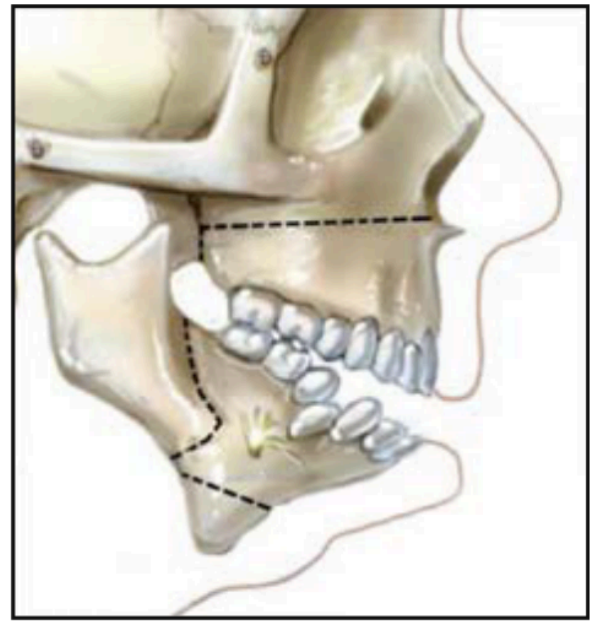


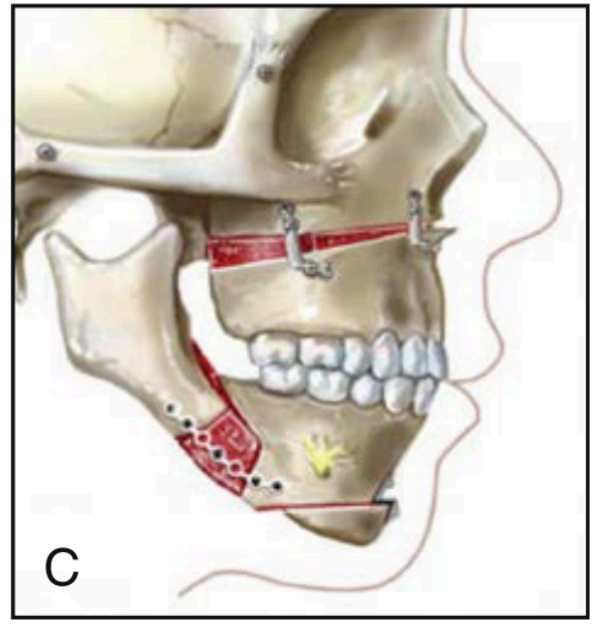
TCS trattamento

1. Regioni orbitaria e malare (innesti)
2. Regione maxillomandibolare (Le Fort 1, osteotomie sagittali mandibola)
3. Regione nasale (rinoplastica)
4. Tessuti molli
5. Orecchio esterno (ricostruzione con innesto di cartilagine)
6. Meato acustico
7. Orecchio medio (ricostruzione catena ossicini o impianto cocleare)



B





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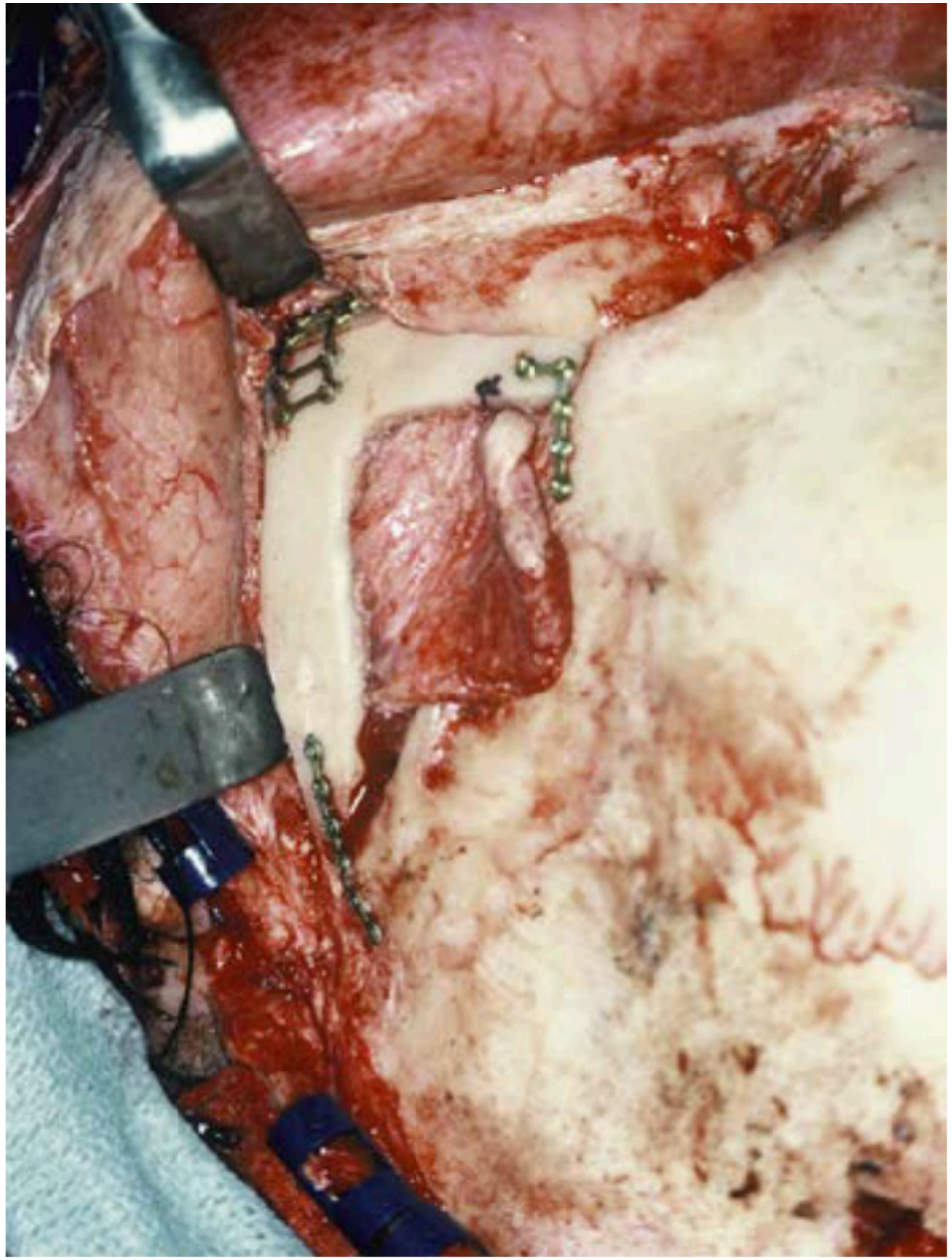
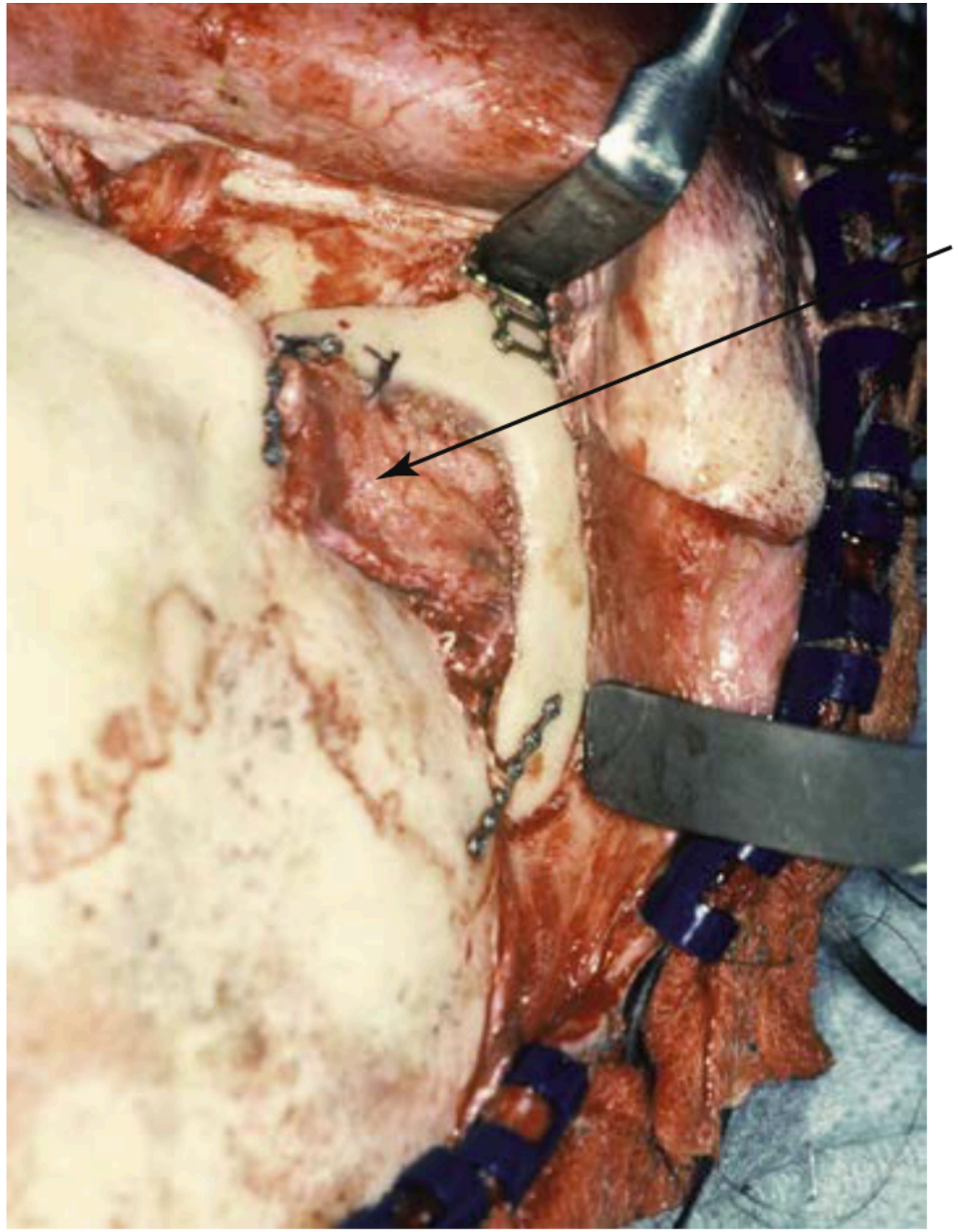
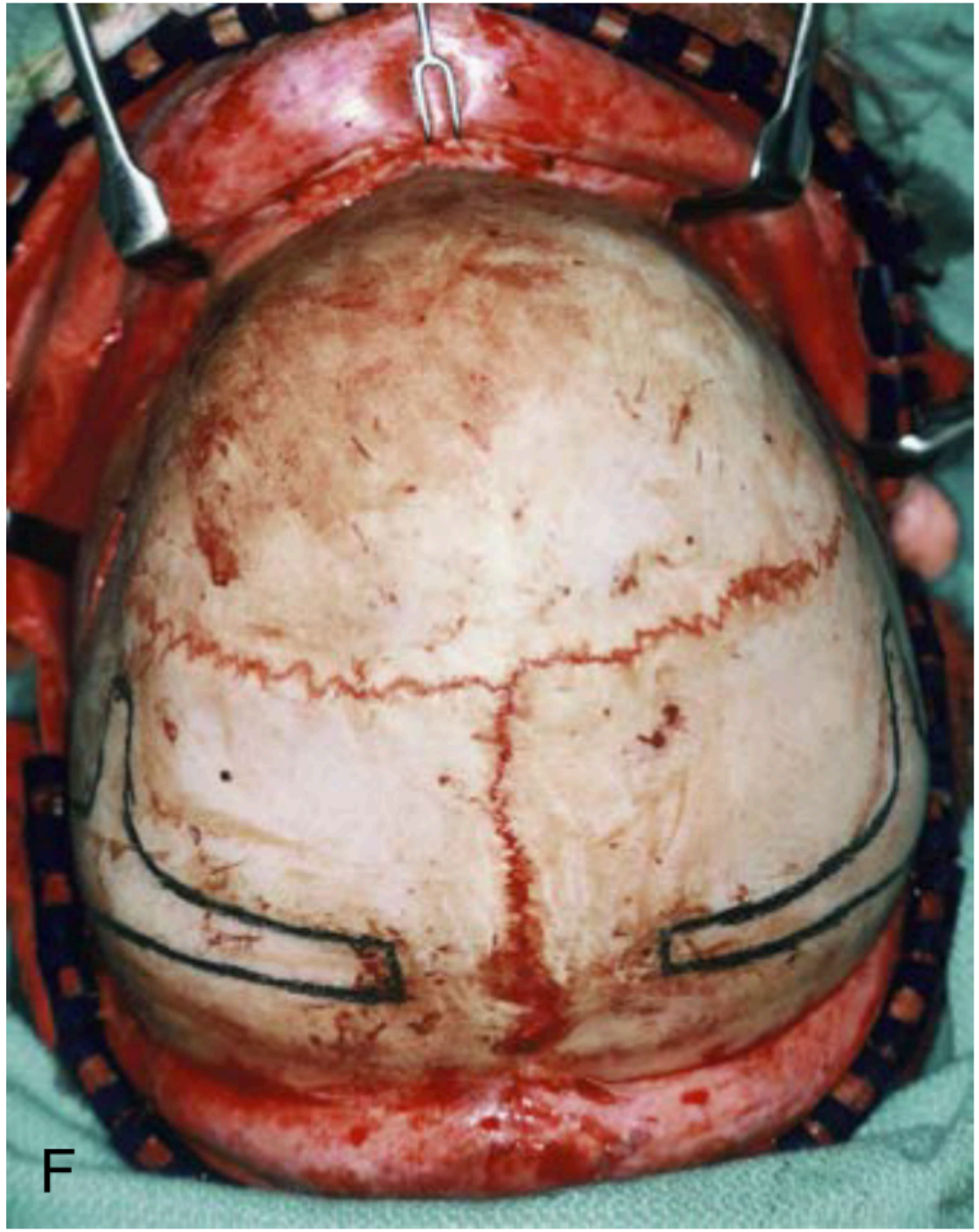
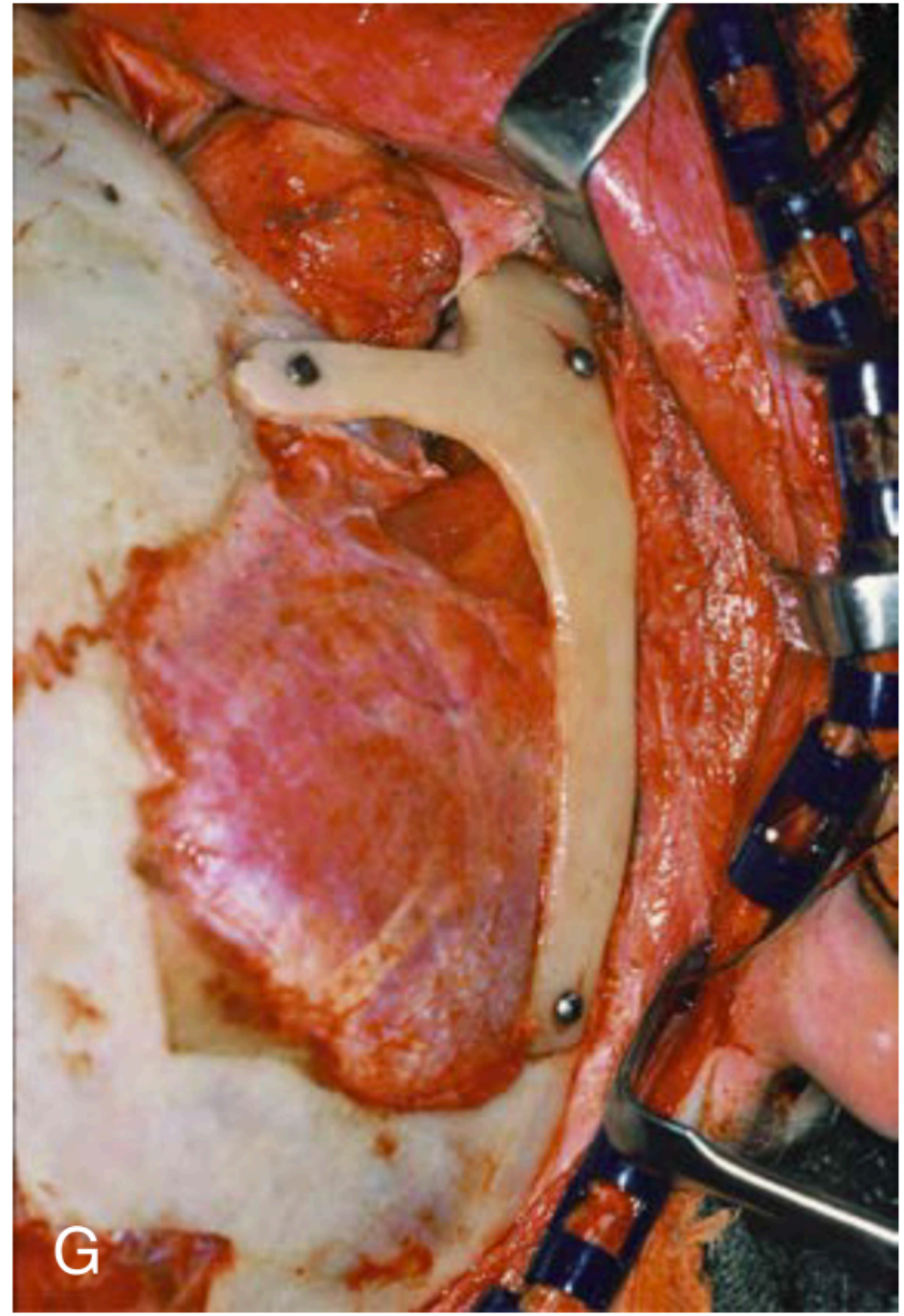
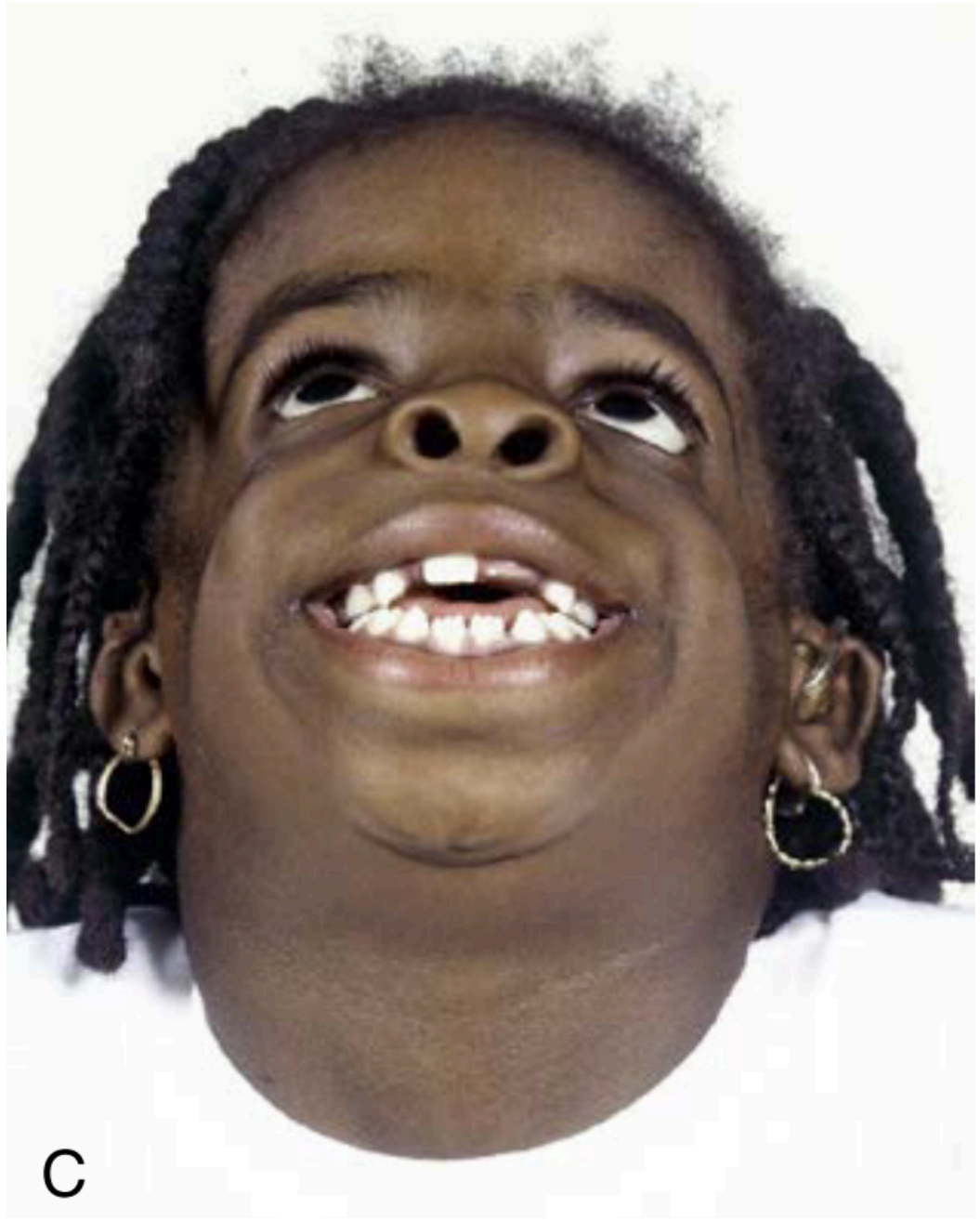



Hypoplastic
temporalis
muscle


F


G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