

Service de chirurgie maxillo-faciale
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Département universitaire
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Rare diseases of calcified tissues of the head and neck

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Centre Hospitalier Régional
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Université Lille 2
Droit et Santé



Inserm

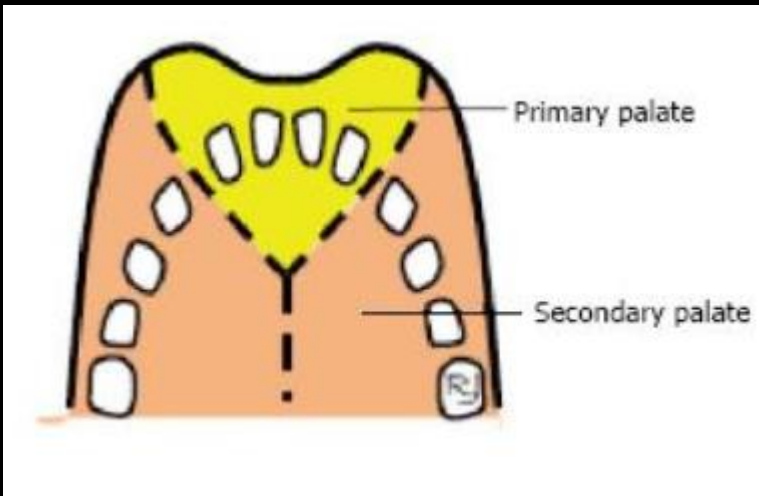
Institut national
de la santé et de la recherche médicale

Why this entity of the : « calcified
tissues of the head and neck »

Embryology/anatomy entity

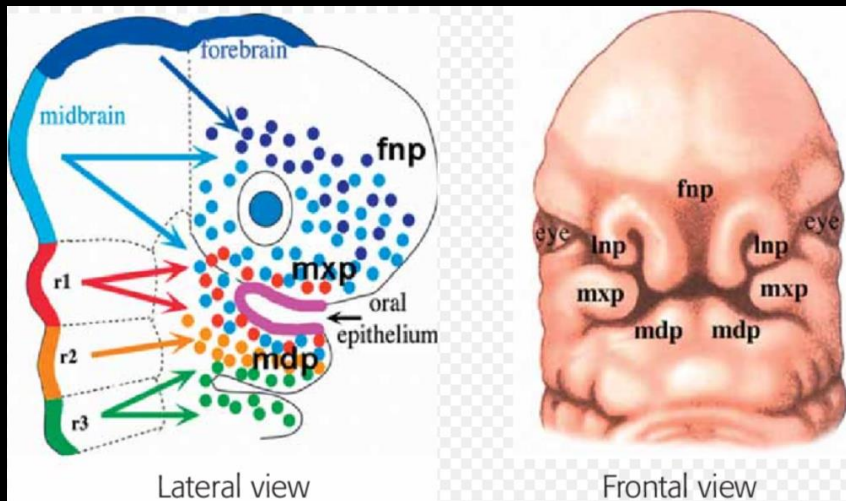


- From cervico-facial arches the calcified tissues of the head and neck are built



The neural crest : The common origin

- The neural crest cells are at the origin of a large part of the cranio-maxillo-facial bones and teeth.



Importance of the genes

From M.C. Meikle

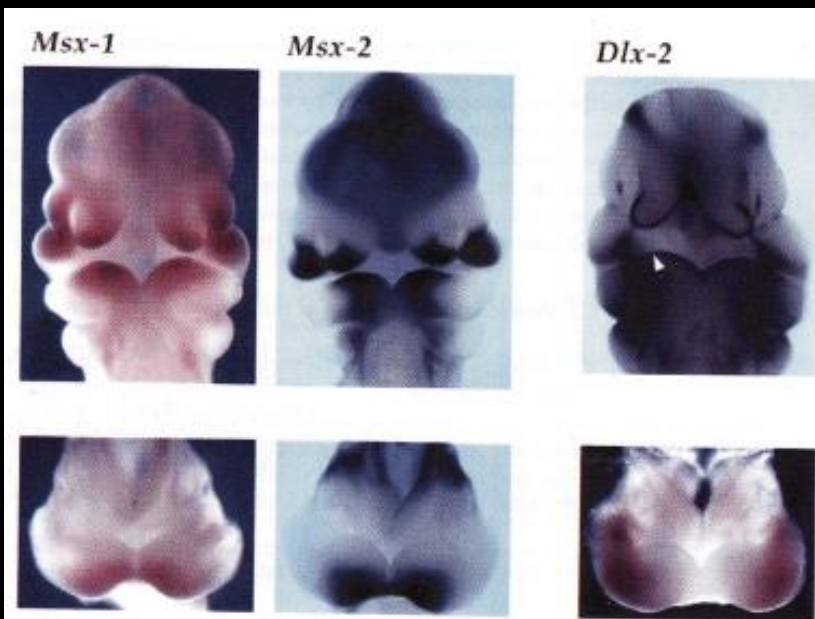


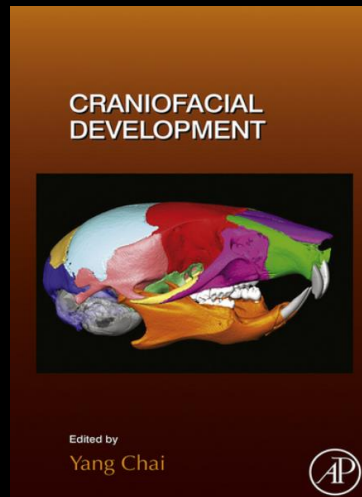
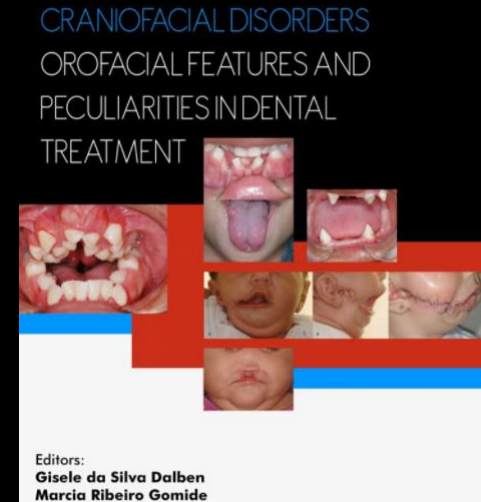
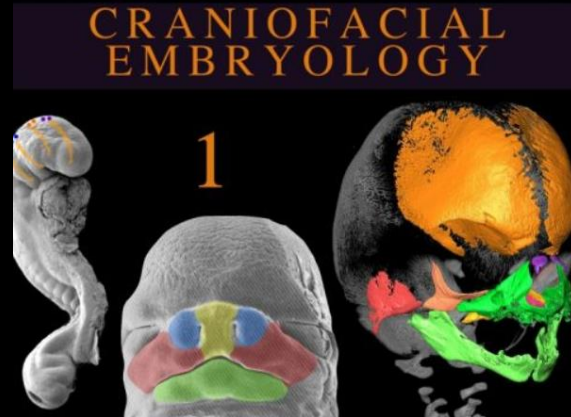
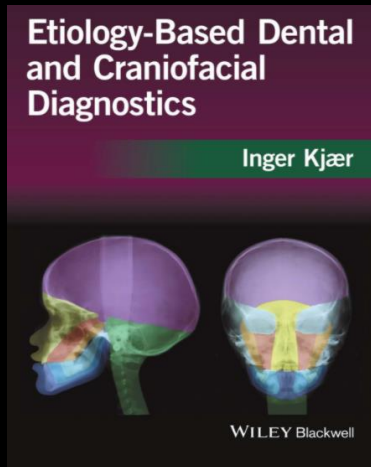
Figure 1.20. *In situ* hybridization of 10.5 day post-conception mouse embryos showing expression patterns of *Msx-1* and *Msx-2* in the distal (ventral) regions of the maxillary and mandibular processes of the first branchial arch, and the medial and lateral nasal processes, and the *Dlx-2* expression in mesenchyme proximally (dorsal) in both the maxilla and mandible and the epithelium distally in the mandible and medial nasal processes. The arrowhead indicates the sharp boundary of expression between the mesenchyme and epithelium. Top, viewed from the ventral aspect; bottom, the mandible removed and viewed from the oral aspect. Whole mount *in situ* hybridization using digoxigenin-labelled RNA probes. [Figures kindly supplied by Dr Bethan L. Thomas.]

- Genes activities
- There are genes involved in the cranio-facial edification and teeth too.

Hormonal/metabolic common impact on craniofacial skeleton and teeth

- Osteogenesis Imperfecta
- Phosphocalcic metabolism
- Etc.

Books, publications, etc.



Etc.

Publications

- **Molecular control of facial morphology**
- [B Liu¹, S M Rooker, J A Helms](#)
- Semin Cell Dev Biol 2010 May;21(3):309-13.
- doi:10.1016/j.semcdb.2009.09.002. Epub 2009 Sep 10.

- **The cephalic neural crest exerts a critical effect on forebrain and midbrain development**
- [Sophie E Creuzet¹, Salvador Martinez, Nicole M Le Douarin](#)
- Proc Natl Acad Sci U S A
- . 2006 Sep 19;103(38):14033-8.
- doi: 10.1073/pnas.0605899103. Epub 2006 Sep 11.

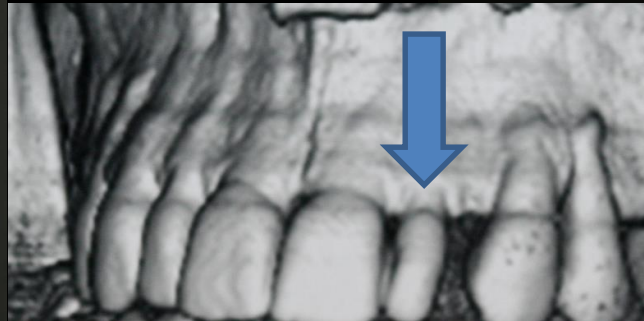
- **Morphoregulation of teeth: modulating the number, size, shape and differentiation by tuning Bmp activity**
- [Maksim V Plikus¹, Maggie Zeichner-David, Julie-Ann Mayer, Julia Reyna, Pablo Bringas, J G M Thewissen, Malcolm L Snead, Yang Chai, Cheng-Ming Chuong](#)
- Evol Dev
- . Sep-Oct 2005;7(5):440-57.
- doi: 10.1111/j.1525-142X.2005.05048.x.

- **The cutting-edge of mammalian development; how the embryo makes teeth**
- [Abigail Tucker¹, Paul Sharpe](#)
- Nat Rev Genet
- . 2004 Jul;5(7):499-508.
- doi: 10.1038/nrg1380.

- Etc.

Examples

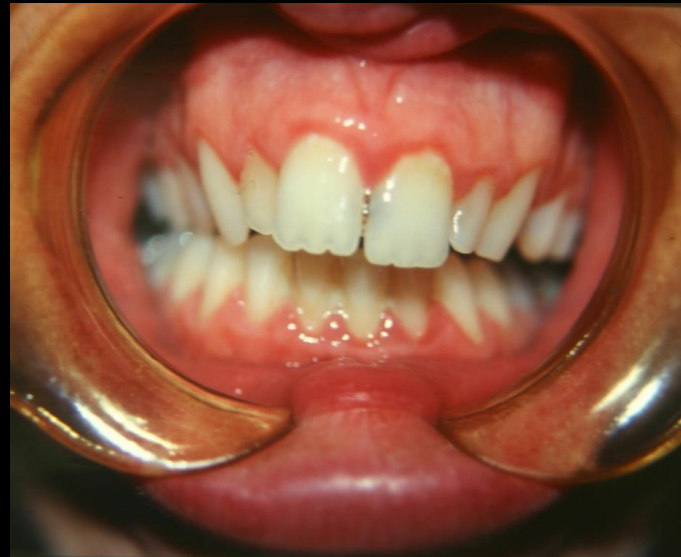
Binder's syndrome



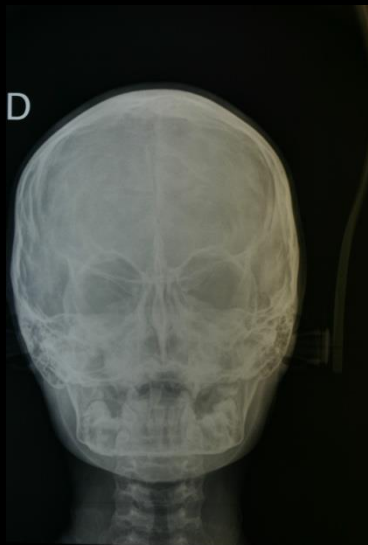
Osteogena imperfecta : (many categories etc.)



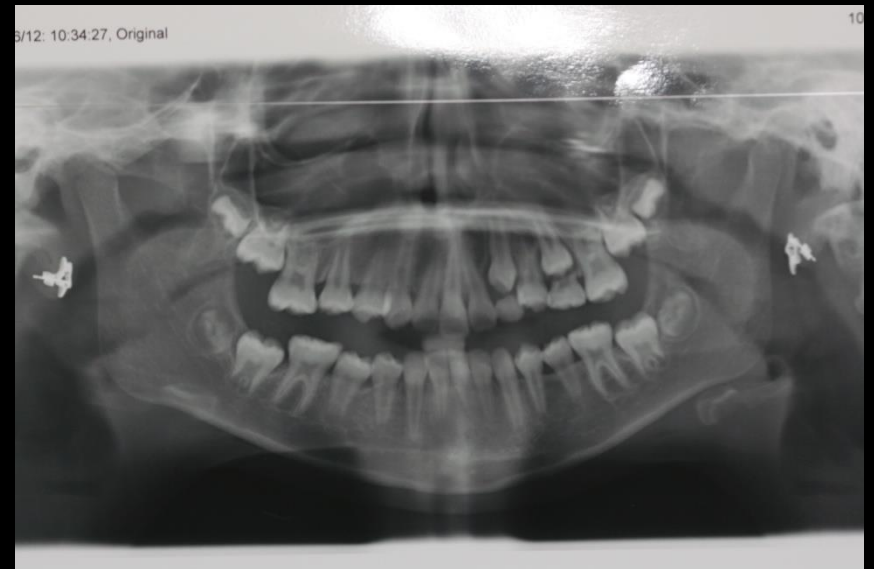
William Beuren's syndrome : Classe II malocclusion, many other features.



Cranio-metaphyseal syndrome



Unique central incisor



Melnick Needles' s syndrome



Cleido-cranial dysostosis



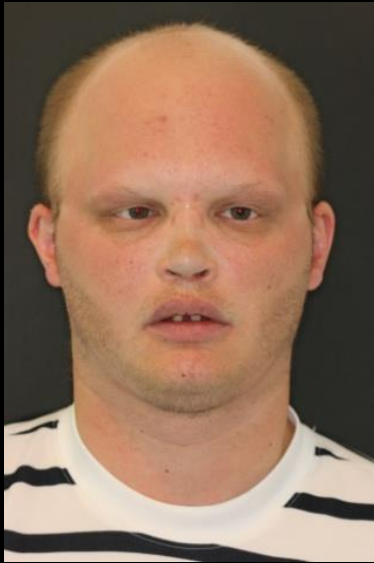
Lacrimo-auriculo-Dento-Digital syndrome (LADD syndrome, Levy Hollister) :



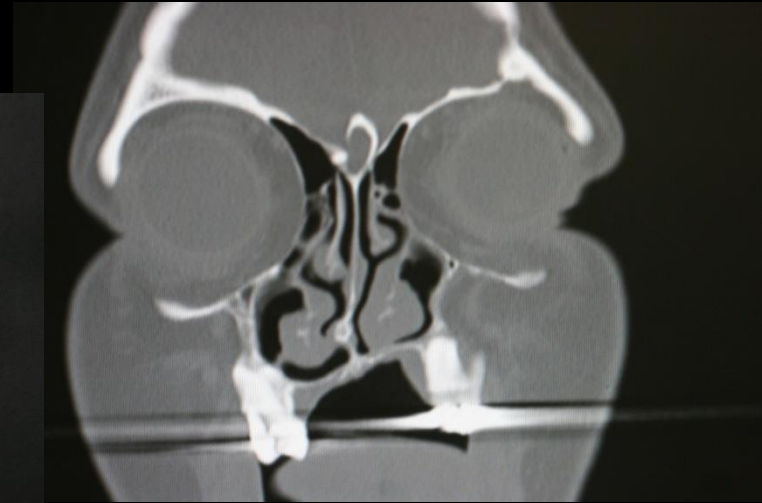
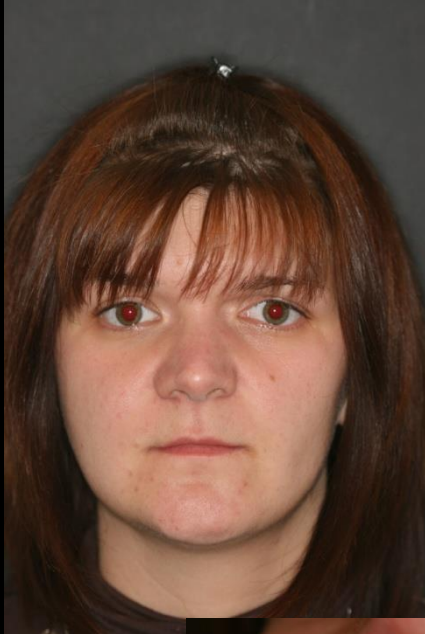
Noonan's syndrome :



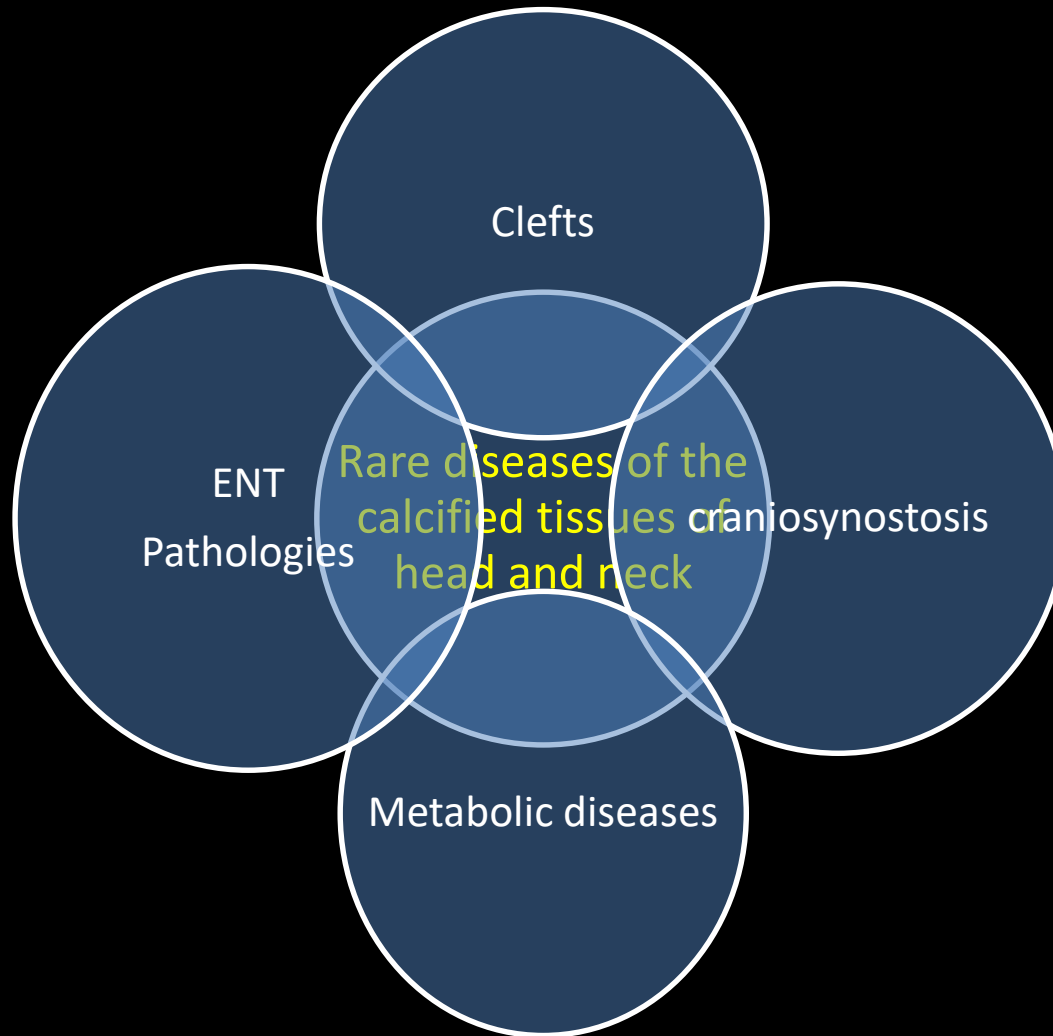
Ectodermal dysplasia (Christ Siemens Touraine syndrome)



Cemento-osseous dysplasia



Position of rare diseases of calcified tissues of the head and neck in the rare diseases group



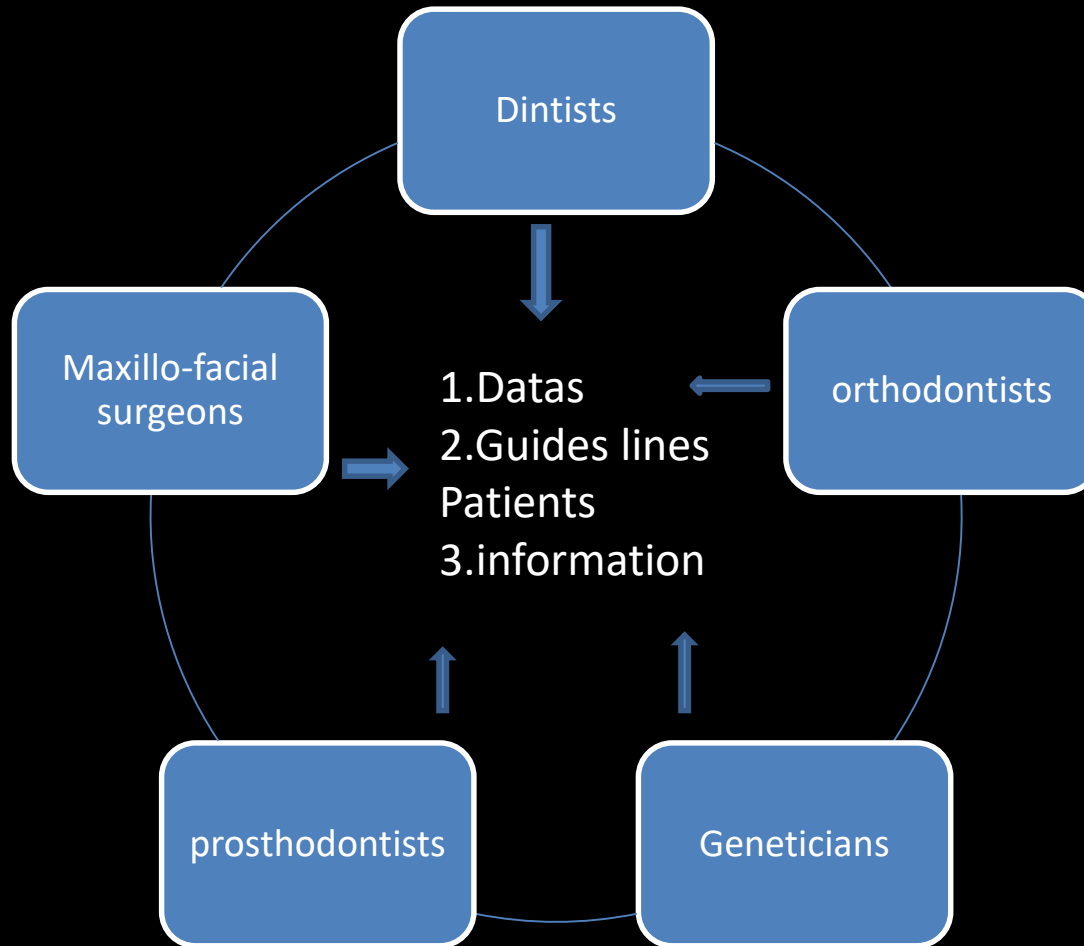
Why is there a necessity to create a group of practitioners involved in these pathologies?

- Numbers of patients
- Unrecognized syndromatic patients
- The absence of coordination of the treatments

Suggestion

- Working group
 - Maxillo-facial surgeons
 - Orthodontist
 - Pediatricians
 - Prosthodontic
 - Geneticians
 - Patients associations

Suggestion : working group



The Working group

- Europe
 - Maxillo-facial surgeons
 - Orthodontists
 - Geneticists
 - Prosthodontists
- North America
 - Orthodontists
- South America
 - Maxillofacial surgeon
- China
 - Maxillo-facial surgeon
- India
 - Maxillo-facial surgeon

Thank you for your kind attention

