J Oral Med Oral Surg 2025;31:E4
https://doi.org/10.1051/mbcb/2025037
© The authors, 2025
https://www.jomos.org

JOMOS
Journal of Oral Medicine and Oral Surgery

Editorial

Rare diseases of the head and neck: the essential role of oral and maxillofacial surgery

Joël Ferri*

Head of the department of Stomatology and Oral and Maxillofacial surgery, Lille University Hospital, France

(Received: 2 July 2025, accepted: 1 October 2025)

For several years, rare diseases, following powerful global lobbying, have established themselves as a clear entity. It was in the United States, driven by patient associations, that the concept of rare diseases was born [1].

Within these rare diseases (which should, moreover, be named under the term of rare or undiagnosed diseases), several medical entities have been identified and then grouped into major categories, sometimes in surprising ways and not always in a scientific logic…

Nevertheless, several groups are rather uncontroversial entities (rare muscle diseases, rare bone diseases, *etc.*). Thus, rare head and neck diseases are a very specific entity with obvious specific clinical manifestations. Moreover, in this region, it is important to remind the frequency of these pathologies and malformations. In addition to its common topographical characteristics, the head entity has an undeniable common embryological and genetic specificity, largely explained by a tissue structure derived from neural crests [2]. These cells, with their immense properties, migrate to numerous cephalic territories and participate in the formation of many craniomaxillofacial structures (including teeth and the craniomaxillofacial skeleton), structures in which our specialty has a clear role to play.

This topographical, genetic, and embryological entity of the head has been identified in several countries, with a "head and neck" network of rare diseases that links together several cephalic specialties [3].

While it is undeniable that each cephalic specialty has a role to play in rare diseases of this region, oral and maxillofacial surgery holds a special and essential role. Indeed, it is important to remember that practitioners specializing in these pathologies must have a strong knowledge of craniomaxillofacial growth, teeth, and their alterations (so common in these diseases). They must also be familiar with surgical techniques for repairing craniomaxillofacial and dental structures [4].

This need of broad knowledge, combining dentistry, medicine, and surgery, that is present in oral and maxillofacial surgeons, who, as a result, can comprehensively understand rare diseases of the head.

If there is one area where oral and maxillofacial surgery is indisputable, it is that of rare head diseases, whose anatomical, embryological, and pathophysiological uniqueness is evident.

Let us hope that our specialty will thus be recognized everywhere as essential in both the diagnosis and treatment of rare head diseases.

References

1. Pearce DA. The history of the international rare diseases research consortium (IRDiRC) and its conferences. Eur J Med Genet. 2024;70:104935.
2. Le Douarin N, Kalchei C. The neural crest. Developmental cell and Biology series, 2nd ed. Cambridge university Press, 1999. ISBN: 0 521 62010 4.
3. Diseases covered by the TETECOU network. https://www.tete-cou.fr/ (consulted 30 June 2025).
4. Kolokythas A. What is the scope of practice of oral and maxillofacial surgeons? J Oral Maxillofac Surg 2021;79:267.

Cite this article as: Ferri J. 2025. Rare diseases of the head and neck: the essential role of oral and maxillofacial surgery. *J Oral Med Oral Surg*. 31: E4. https://doi.org/10.1051/mbcb/2025037

* Correspondence: ferri.joel@gmail.com

This is an Open Access article distributed under the terms of the Creative Commons Attribution License (https://creativecommons.org/licenses/by/4.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.