Working Group

of

Rare Diseases of the Boning Tissues of the Head

(WG RDBTH)

Introduction

Rare diseases of the head are involving a considerable number of pathologies. Several explanations can be given for this high number of diseases:

1 The variety of tissues that are present in this region. Therefore, any impairment of the fundamental tissues has an expressivity in the cephalic area.

2 A certain embryological complexity with contiguity of the sensorial organs (optical, otic, olfactory and auditory placodes) and of the central nervous system.

3 Specific tissues which all originate at different degrees from the neural crests cells.

With regard to Boning tissues, there is added to the causes already mentioned, a particular ossification (membranous ossification), which moreover is sensitive to any general anomaly of ossification with consequences on the cranio-maxillo-facial skeleton.

Morever, bone and tissues related to them via their embryology origin, as boning tissues of the cephalic extremity, have a unique embryological history, which explains a specific "behaviour" in particular pathological situations. At the genetic level there are a large number of genes involved in the edification of the cranio-maxillo-facial skeleton and tissues related to it. Finally, the variety of potential defects also explains why the signs of these defects are sometimes “minor”, which induces late diagnoses or unrecognized pathologies.

Several publications demonstrate this (1,2, 3,4,5).

Patients with these pathologies require an accurate diagnosis and specific care. There often need specific care including early orthodontics, pre-prosthetic, prosthetic, implant and surgical treatments.

The whole treatment is often quite specific to oral and maxillofacial surgery.

Therefore there is an interest in thinking about this entity of RDBTH that includes specific and rare pathologies and that are frequently underdiagnosed.

The creation of the RDBTH group within the MJC RUD is therefore essential.

Pathologies covered by RDBTH spectrum

It is difficult to describe all the pathologies that have manifestations on the boning tissues of the cephalic extremity.

However we can suggest 5 groups of pathologies.

1 Specific syndromes (Cleidocranial dysostosis, Rubinstein Taybi, Corneila Delange, Binder, Pyle, Melnick-needles, Levy-Hollister, Noonan, Cherubinism, condylo-mandibulo dysplasia, Incontinenta Pigmenti, Kenny Caffey, etc…). Sometimes these syndromes are known, sometimes there are not, even if the clinician sees that there is an "abnormality", frequently he is not able to recognize it. Above all, there is no coordination in the management of these patients who are often lost without being informed of the way to be properly treated.

2 Disorders of calcified tissue edification. This group includes osteogena Imperfecta associated with amelogena and/or dentinogena imperfecta, amelo-dentinogena imperfecta, lacrimo auriculo-dento-digital syndrome,picnodysostosis, syndromic and non-syndromic dental agenesis,etc. These pathologies are related to disorder of the boning tissues edification. Unfortunately, there is no coordination of the dento-maxillo-facial treatments.

3 Ectodermal dysplasias that most of time have dento-maxillo-facial defects. More largely in this group we can include all the dento-bony dysplasias.

4 Localized bone dysplasias (fronto-metaphyseal dysplasias, localized anomalies of dento-osseous structures, etc.)

5 Specific syndromes affecting the bones of the maxilla-facial structure (cemento-osseous dysplasia, primary failure of tooth eruption).

RDBTH Working Group structure

The UEMS MJC RUD RDBTH WG is the coordinator, and will act as a steering comity (board of directors). It will include at least one oral and maxillofacial surgeon, one orthodontist, one paediatrician, one geneticist (Suggestion of the

persons that could be immediately in the board) : J Ferri, S Politis, R Radalanski/ T Koehne, B Melegh, L Rasmusson, R Kontio, Z Nemeth, Paediatrician?)

Because of the diversity of the specialities that are involved in the RDBTH it is mandatory to have these specialists in the Board. This board of directors is elected among the members of the group. It includes a president, a secretary, and **advisors (dedicated to relations with the other sections involved in the RUD and/or other specialists**). The president is in charge of the coordination of the group.

This RDBTH working group is open to any specialist whatever his nationality and speciality **provided that he (or she) has demonstrated activities in diagnostic or treatment of RDBTH and provided that there are validated by the UEMS MJC RUD RDBTH**. Admission is validated by the board of directors after checking the CV of the candidates. Candidatures must be supported by two godfathers.

There is at least one meeting/year (general assembly). During this meeting the board of directors is deciding action and refer its activities to the MJC RUD.

FUTURE DIRECTIONS

At the national level (one representative / nation involved). Ideally, there should be representatives for each European country. Maximum 4 representatives/ country will be accepted (ideally belonging to specialties of Oral and maxilla-facial surgery, orthodontics, Geneticist, prosthodontist).

At the international level the rules will be the same. Here are the suggested candidates that have confirmed their participation with selected colleagues from their country. This will be done under the umbrella of the European Board of Rare and Undiagnosed Diseases.

Proposed actions of RDBTH

An annual meeting will be set up with three lines of work.

1 Pooling of scientific data

The pooling of scientific data is one of the concerns of our RDBTH group. These scientific exchanges take the form of congresses or scientific meetings. During these meetings the latest scientific data are exposed and discussed and validated or not. This meeting is set up by the steering comity.

The establishment of therapeutic guidelines. The implementation of the guidelines is mandatory for the practitioners taking in charge these pathologies. They help colleagues to recognize difficult-to-diagnose pathologies and, once the diagnosis has been done, to propose appropriated treatments. The guidelines are also set up for

patients or families of patients in order to guide them in the management of pathologies which sometimes require several areas of expertise.

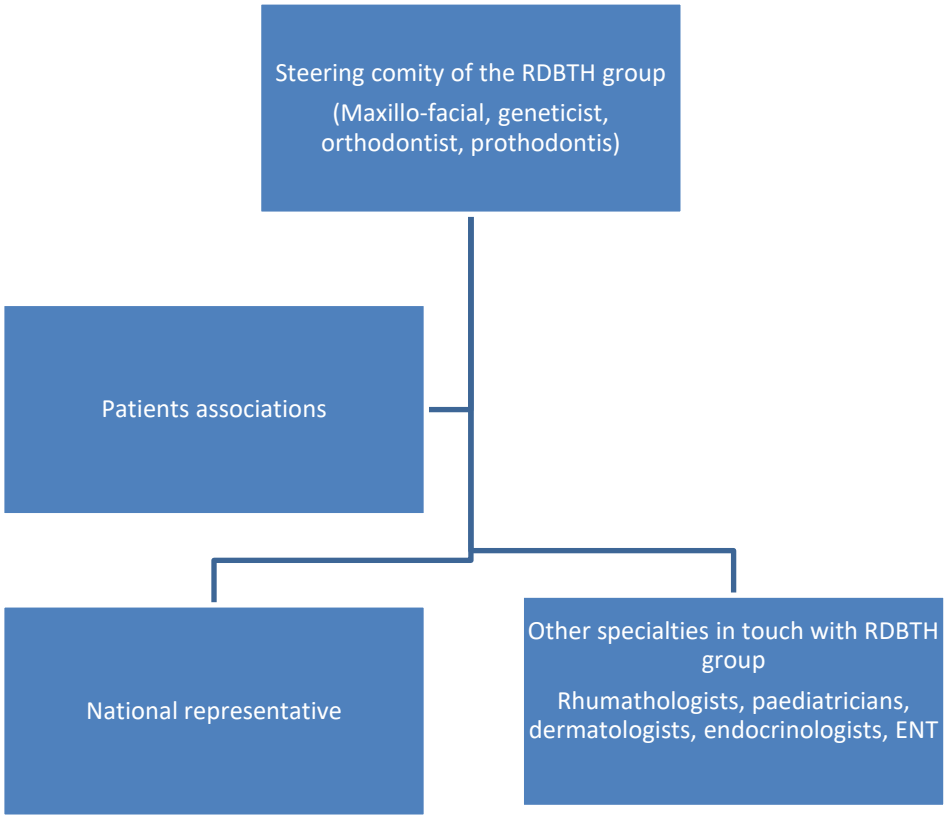
2 Actions with associations

Information from scientific data is fed back to patients through an annual meeting. This meeting is necessary in order to allow a clear transmission of information. These relationships with patient associations also make it possible to develop the best strategies for disseminating information to their members but also more widely to patients and the population. Patient associations must therefore be involved in the actions to be taken.

3 Relations with other specialties

Some specialties can face rare diseases of the calcified tissues of the head (dermatologists, rheumatologists, pediatricians, endocrinologists, ENT). Furthermore, these specialties may have to deal with certain manifestations of these diseases, as far as their field is concerned. It is therefore important that these specialties can have contact with our group

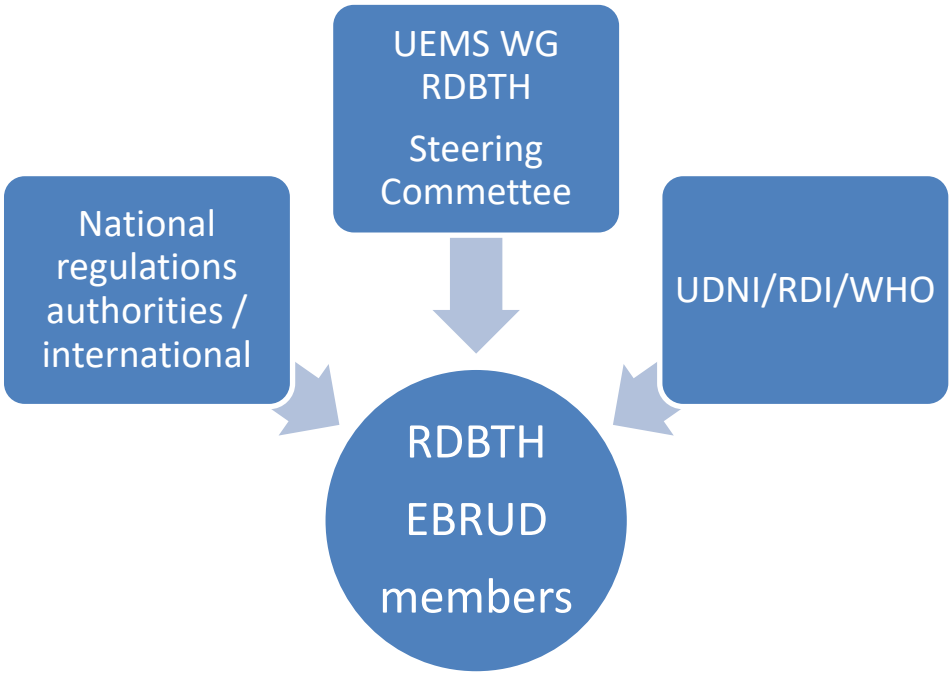
Example: One can cite as example dermatology which often encounters ectodermal dysplasia but does not deal with reconstructions of the jaws or teeth.



Relation with public health authorities and institutions

The group follows the international rules of the association of rare diseases and maintains contact with public authorities at national and international level.

Our group plans to stay close to the Undiagnosed Diseases Network International (UDNI) in order to actively participate at scientific meetings and suggest actions. The UDNI is the reference structure for rare diseases, it seems essential for us to participate regularly in its scientific events. Our group also wishes to be in touch with the network “Rare Diseases International (RDI)”, and “World Health Organization (WHO)” which has a sector supporting rare diseases.



Participation at the EBRUD

The RDC group intend to participate at the EBRUD in order to implement this exam in order to point out the importance of these pathologies that represent a large spectrum of pathologies actually frequently ignored.

Références

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