ROMSE- A database for orofacial manifestations in patients with rare diseases and setup of a unified classification form of dysgnathia

Hanisch L.[1], Hanisch M.[2], Kleinheinz J.[2], Jackowski J.[3], Danesh G.[1]

[1] Department of Orthodontics
Faculty of Health, School of Dentistry.
Witten/Herdecke University.
Alfred-Herrhausen-Str. 44
58455 Witten, Germany.

[2] Department of Cranio-Maxillofacial Surgery
University Hospital Münster
Albert-Schweitzer-Campus 1
48149 Münster, Germany

[3] Department of Oral Surgery and Dental Emergency Care
Faculty of Health, Witten/Herdecke University
Alfred-Herrhausen-Strasse 45
58455 Witten, Germany

Introduction

Around 30 million people in Europe are affected by a rare disease. Approximately 80% of all known rare diseases are of genetic origin and around 15% can become manifest in the orofacial region[1,2], e.g. craniofacial dysplasia such as cleft lip and palate, dysgnathia, and hypodontia. Orthodontics forms a major field in rare diseases, and orthodontists are often the first ones to come in contact with young patients who are affected by a rare disease. There is little knowledge in dentistry about how to treat patients with rare diseases and that an orofacial manifestation can help to find the diagnosis. Our aim is to establish a "database for orofacial manifestations in people with rare diseases-ROMSE“ [3] and to concentrate the little knowledge on rare diseases in order to improve the diagnosis and treatment of patients with rare diseases. To allow a standardised documentation of orthodontic cases, it is necessary to unify the classifications of dysgnathia.

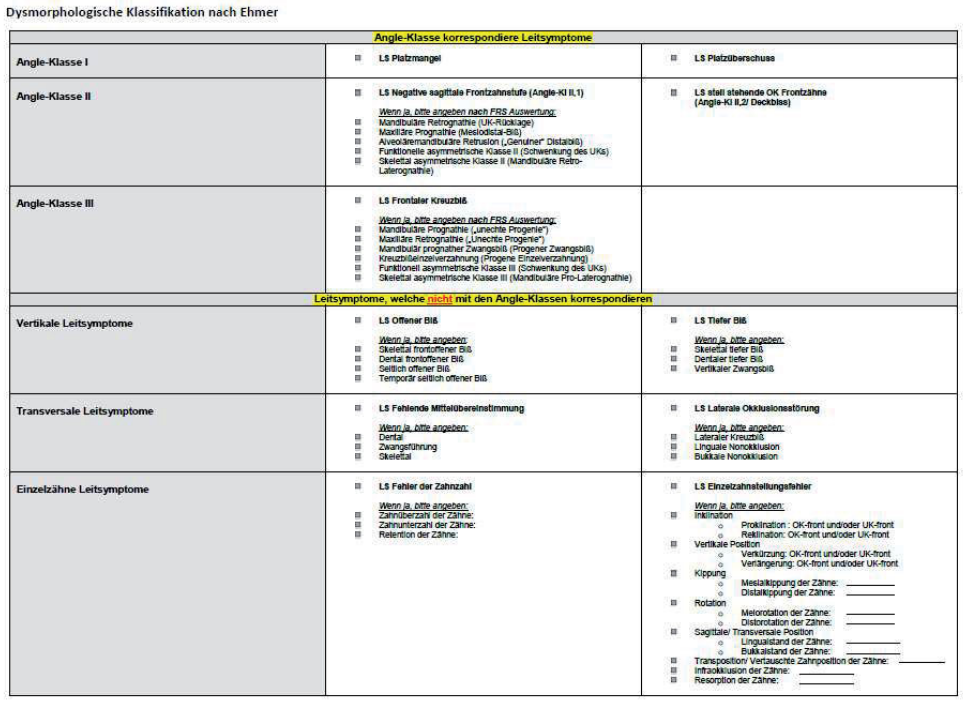
Material and Methods

Since 2011 material from various databases like Orphanet, OMIM, and Pubmed, was evaluated. Starting in 2013 the gathered information has been incorporated into a web-based, freely accessible database at http://romse.org. The dysmorphological classification of “Ehmer” [4] shall be the guideline for orthodontists to classify the dysgnathia and to standardise the documentation of people with rare diseases. The classification form is freely available at the ROMSE website.

Results

So far 531 rare diseases with orofacial manifestations have been listed in the ROMSE database. Up to now, 10 global categories for orofacial manifestations such as dental anomalies, dysgnathia or orofacial clefts which are subdivided into 100 subcategories have been set up.. About one third of those diseases or syndromes show dysgnathia. Especially the sub-classification of dysgnathia seems to be difficult since most of the patients were not analysed according to a standardised classification. Wrong or double assignments are the result. To unify the classifications of dysgnathia, a modified version of the “Dysmorphological Classification by Ehmer“ adapted for rare diseases was developed (Fig. 1).

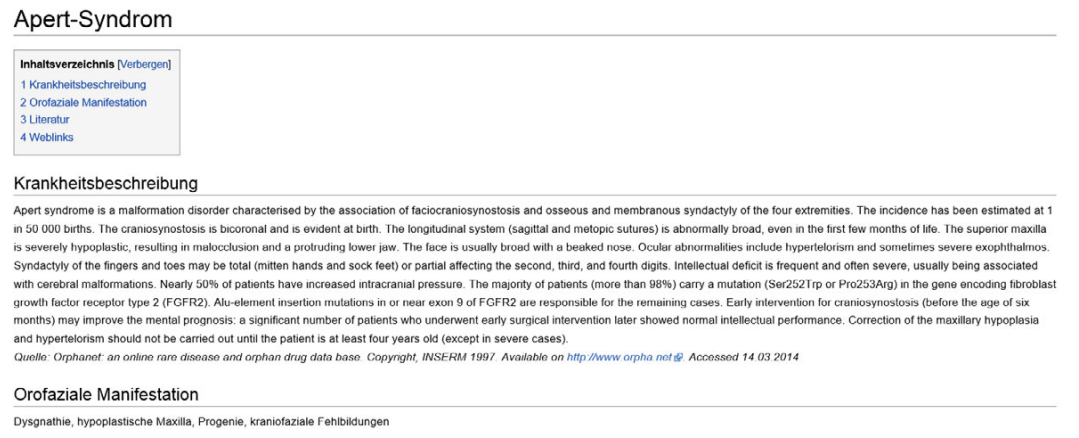
Figures



Dysmorphologische Klassifikation nach Ehmer

	Angle-Klasse korrespondiere Leitsymptome	
Angle-Klasse I	▪ LS Platzmangel	▪ LS Platzüberschuss
Angle-Klasse II	▪ LS Negative sagittale Frontzahnstufe (Angle-Kl II,1) *Wenn ja, bitte angeben nach FRS Auswertung:* ▪ Mandibuläre Retrognathie (UK-Rücklage) ▪ Maxilläre Prognathie (Mesialstar-Biß) ▪ Alveolärer mandibuläre Retrusion („Genuiner“ Distalbiß) ▪ Funktionelle asymmetrische Klasse II (Schwenkung des UKs) ▪ Skelettal asymmetrische Klasse II (Mandibuläre Retro-Laterognathie)	▪ LS steil stehende OK Frontzähne (Angle-Kl II,2/ Deckbiss)
Angle-Klasse III	▪ LS Frontaler Kreuzbiß *Wenn ja, bitte angeben nach FRS Auswertung:* ▪ Mandibuläre Prognathie („unechte Progenie“) ▪ Maxilläre Retrognathie („unechte Progenie“) ▪ Mandibulär prognather Zwangsbiß (Progener Zwangsbiß) ▪ Kreuzbißeinzelverzahnung (Progene Einzelverzahnung) ▪ Funktionell asymmetrische Klasse III (Schwenkung des UKs) ▪ Skelettal asymmetrische Klasse III (Mandibuläre Pro-Laterognathie)	
	Leitsymptome, welche nicht mit den Angle-Klassen korrespondieren	
Vertikale Leitsymptome	▪ LS Offener Biß *Wenn ja, bitte angeben:* ▪ Skelettal frontoffener Biß ▪ Dental frontoffener Biß ▪ Seitlich offener Biß ▪ Temporär seitlich offener Biß	▪ LS Tiefer Biß *Wenn ja, bitte angeben:* ▪ Skelettal tiefer Biß ▪ Dentaler tiefer Biß ▪ Vertikaler Zwangsbiß
Transversale Leitsymptome	▪ LS Fehlende Mittelübereinstimmung *Wenn ja, bitte angeben:* ▪ Dental ▪ Zwangsführung ▪ Skelettal	▪ LS Laterale Okklusionsstörung *Wenn ja, bitte angeben:* ▪ Lateraler Kreuzbiß ▪ Linguale Nonokklusion ▪ Bukkale Nonokklusion
Einzelzähne Leitsymptome	▪ LS Fehler der Zahnzahl *Wenn ja, bitte angeben:* ▪ Zahnüberzahl der Zähne: ▪ Zahnunterzahl der Zähne: ▪ Retention der Zähne:	▪ LS Einzelzahnstellungsfehler *Wenn ja, bitte angeben:* ▪ Inklination ○ Proklination : OK-front und/oder UK-front ○ Reklination: OK-front und/oder UK-front ▪ Vertikale Position ○ Verkürzung: OK-front und/oder UK-front ○ Verlängerung: OK-front und/oder UK-front ▪ Klippung ○ Mesialklippung der Zähne: _____ ○ Distalklippung der Zähne: _____ ▪ Rotation ○ Mesiorotation der Zähne: _____ ○ Distorotation der Zähne: _____ ▪ Sagittale/ Transversale Position: ○ Lingualstand der Zähne: _____ ○ Bukkalstand der Zähne: _____ ▪ Transposition/ Vertauschte Zahnposition der Zähne: _____ ▪ Infraokklusion der Zähne: _____ ▪ Resorption der Zähne: _____

Fig. 1: Modified version of the “Dysmorphological Classification by Ehmer“



Apert-Syndrom

Inhaltsverzeichnis [Verbergen]
1 Krankheitsbeschreibung
2 Orofaziale Manifestation
3 Literatur
4 Weblinks

Krankheitsbeschreibung

Apert syndrome is a malformation disorder characterised by the association of faciocraniosynostosis and osseous and membranous syndactyly of the four extremities. The incidence has been estimated at 1 in 50 000 births. The craniosynostosis is bicoronal and is evident at birth. The longitudinal system (sagittal and metopic sutures) is abnormally broad, even in the first few months of life. The superior maxilla is severely hypoplastic, resulting in malocclusion and a protruding lower jaw. The face is usually broad with a beaked nose. Ocular abnormalities include hypertelorism and sometimes severe exophthalmos. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. Nearly 50% of patients have increased intracranial pressure. The majority of patients (more than 98%) carry a mutation (Ser252Trp or Pro253Arg) in the gene encoding fibroblast growth factor receptor type 2 (FGFR2). Alu-element insertion mutations in or near exon 9 of FGFR2 are responsible for the remaining cases. Early intervention for craniosynostosis (before the age of six months) may improve the mental prognosis: a significant number of patients who underwent early surgical intervention later showed normal intellectual performance. Correction of the maxillary hypoplasia and hypertelorism should not be carried out until the patient is at least four years old (except in severe cases).

Quelle: Orphanet: an online rare disease and orphan drug data base. Copyright, INSERM 1997. Available on http://www.orpha.net. Accessed 14.03.2014

Orofaziale Manifestation

Dysgnathie, hypoplastische Maxilla, Progenie, kraniofaziale Fehlbildungen

Fig. 2: Selected example: “Apert Syndrome“ listed in the ROMSE database

Literatur

- PMID 23614948
- PMID 22449603
- PMID 20809043

Weblinks

- http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=87
- http://omim.org/entry/101200

Kategorien: Dysgnathie | Kraniofaziale Fehlbildung | Maxilla, hypoplastisch | Progenie

Fig. 3: Selected example: “Apert Syndrome“ listed in the ROMSE database

Kategorie:Dysgnathie

Definition

Alle Fehlentwicklungen der Zähne, der Kiefer und/oder des Kausystems. Die Anomalien können Zahnstellung, Okklusion, Kieferform, die Lage der Kiefer zueinander oder den Einbau der Kiefer in den Schädel betreffen.

Unterkategorien

Es werden 15 von insgesamt 15 Unterkategorien in dieser Kategorie angezeigt.

G
- [×] Gaumen,hoher (33 S)

M
- [×] Makrostomie (6 S)
- [×] Malokklusion (18 S)
- [×] Mandibula,hyperplastisch (1 S)
- [×] Mandibula,hypoplastisch (9 S)

M (Fortsetzung)
- [×] Maxilla, hypoplastisch (24 S)
- [×] Maxilla,hyperplastisch (4 S)
- [×] Maxilla,Retrusion (1 S)
- [×] Mikrogenie (65 S)
- [×] Mikrognathie (17 S)

M (Fortsetzung)
- [×] Mikroretrogenie (4 S)
- [×] Mikrostomie (13 S)

P
- [×] Progenie (22 S)

R
- [×] Retrogenie (5 S)
- [×] Retrognathie (7 S)

Fig. 4: Selected example: “Dysgnathia“ listed in the ROMSE database

Conclusions

Rare diseases and their symptoms come with difficult challenges regarding their therapy. More than 900 rare diseases with a genetic genesis can be demonstrated to have a dental, oral, or maxillofacial involvement [2]. Since publications on rare diseases and data on possible care strategies in dental medicine are scant, there is an urgent need to provide the limited available information on a central and accessible platform. By setting up a "database for orofacial manifestations in people with rare diseases-ROMSE“, a platform is provided for dentists and orthodontists to work interdisciplinarily on treatment strategies [3]. A consistent and beyond dentistry classification of dysgnathia can prospectively avoid wrong assignments.

Contact: Dr. Lale Hanisch, *lale.hanisch@uni-wh.de*

Literature:

1. Jackowski J, Hanisch M. Orofaziale Manifestationen bei 2006 seltenen Erkrankungen- ein vorläufiger systematischer Literaturreview. Deutsche Zahnärztliche Zeitschrift 2012; 67(6).
2. John Hopkins University: OMIM – Online Catalog of Human Genes and Genetic Disorders. [www.ncbi.nlm.nih.gov/omim] accessed 22.01.2015.
3. Hanisch M, Hanisch L, Benz K, Kleinheinz J, Jackowski J. Development of a database to record orofacial manifestations in patients with rare diseases: a status report from the ROMSE (recording of orofacial manifestations in people with rare diseases) database. Br J Oral Maxillofac Surg. 2017, doi: 10.1016/j.bjoms.2017.02.003.
4. Praxis der Zahnheilkund. Kieferorthopädie 1. Hrsg. P. Diederich, 4. Auflage 2000, Urban und Fischer Verlag München, Jena.