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REVIEW ARTICLE

Oral and craniomaxillofacial syndromes and disorders: the person behind the name

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Abstract

Objectives: A syndrome is a correlated set of medical signs and symptoms. This study seeks to develop a non-exhaustive list of biographies of medical eponym. Researchers who originally described and named oral and craniomaxillofacial syndromes and disorders have contributed widely used figures.
Methods: The authors searched Pubmed for names of researchers in quotation marks, one by one. Two websites, a medical name dictionary and a free encyclopedia were taken over to expand the author´s data. The original work and the year of publication were communicated to each author.
Results: A list of 20 biographies was compiled that were relevant to 18 distinct oral and craniomaxillofacial syndromes or disorders was compiled and sorted by year of publication.
Conclusions: The use of medical names is not necessary for anatomical applications alone. Syndromes and disorders are better communicated when medical eponyms are used. The authors believe that namesake on this topic will not stop being used.

Keywords: Biography; Craniofacial abnormalities; Dentofacial deformities; Eponym; Historical Article; History of Dentistry; History of Medicine; Humans; National Library of Medicine; Syndrome

Introduction

The use of medical eponyms was common. There have been reports since the 11th century, when Avicena proposed an idea of a specific disease diagnosis. From the 1970s, there was a recommendation to avoid this practice. The use of medical eponyms poses a drawback to scholar writing and medical education [1]; in addition, some authors blame medical eponyms as a source of confusion among learners [2]. Understandable non-use of eponyms in connection with anatomical accidents. In the case of surgical instruments or techniques [3–5], eponyms make it easier to understand and memorize. The same question can be asked in medical eponyms related to syndromes or disorders. The role of eponyms enables a faster and broader understanding of medical signs and symptoms.

This paper provides some succint biographies of health profesisonals who have identified oral and craniomaxillofacial syndromes and disorders as researchers whose historical contribution to science must never be forgotten.

Materials and methods

The authors compiled a list of eponyms relevant to oral and craniomaxillofacial syndromes and disorders. A bibliographic search was carried out on Pubmed for the names of the researchers in single quotation marks. Two websites have been implemented to achieve some data not available on this bibliographic search in order to expand the data on authors. Whonamedit is a dictionary of medical eponyms [6], and a list of eponymous disorders of the same name at Wikipedia [7], the largest and a free global encyclopedia. Original description and publication of syndromes and disorders were given in each author's biography. Biographies were sorted by year of publication. Pronunciation of names in langagues vastly dissimilar from English has been provided. As a biography, this article is exempt from institutional approval.

Results

Bell´s palsy

Sir Charles Bell (1774-1842) was a Scottish surgeon, anatomist, and artist who first detailed facial paralysis in 1821 [8] (figure 1). Bell´s palsy (figure 2) is a facial paralysis, partially or complete. It increases the susceptibility to noises and changes the taste.

Figure 1.Portrait of Sir Charles Bell by H. Goffey after J. Stevens

Figure 2.Patient with Bell´s palsy

A common ethiology is the reactivation of the herpes virus on facial nerve [9]. He was from Edinburgh and was one of the creators of Middlesex Hospital Medical School. Professor at the College of Surgeons in London, the King´s College of London, and the University of Edinburgh. He was made a knight in 1833.

He should not be confused with Julia Bell (1879-1979), a pioneering English geneticist named Martin-Bell syndrome, known as Fragile X syndrome

Parry-Romberg syndrome

Hemifacial microsomia, Romberg syndrome, or Parry-Romberg syndrome is flagged progressive hemifacial atrophy, limb glitches and neurological manifestations [10].

Caleb Hillier Parry (1755-1822) was a British doctor from Cirencester (figure 3). Although considered a provincial physician, he made notable discoveries, including the bradycardic influence of carotid artery compression and the link between thyroid and heart disease [11]. Friend of Edward Jenner (1749-1823), a pioneer of the vaccine idea. First to mention Parry-Romberg syndrome, was unable to work due to a stroke; his works were published posthumously, in 1825 [12].

Moritz Heinrich Romberg (1795-1873) was a German neurologist, from Meiningen (figure 4).

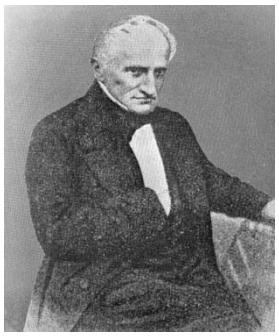
Figure 3 Engraving of Caleb Hillier Parry

Figure 4 Lithograph of Moritz Heinrich Romberg by Wildt after J. Schlesinger

Postgraduated in Vienna, is considered one of the early founders of Neurology [13], a well-known and highly respected teacher. Famous for the Romberg´s sign, which is common to all proprioceptive disorders of the legs [14]. Parry-Romberg syndrome was assessed and discussed, supplemented by the description of progressive hemifacial atrophy, the work of Parry being continued [15].

Ludwig´s angina

Wilhelm Frederick von Ludwig (1790-1865) was a German surgeon and obstetrician, from the Duchy of Wirtemberg, near Stuttgart (figure 5). He is known for his publication in 1836 [16] on a life-threatening illness, Ludwig´s angina, a fast-spreading infection involving fascial spaces [17] as seen in Figure 6.



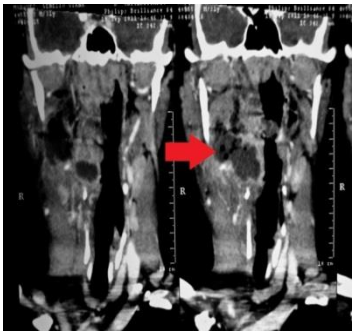


Figure 5. Wilhelm Frederick von Ludwig

Figure 6. A tomographic image that show an infection (red arrow) that is compromising the airway.

He was praised at 19, was a teacher at 25, president of the Württemberg Medical Association and Chief-doctor to the royal family [18]. However, ironically, he could have died of his own condition [19].

Down syndrome

Trisomy 21 or Down syndrome is the very popular genetic disorder in humans, 1/1,000 newborns. Affects many organ systems, including neurodevelopmental disorders, low muscle tone, and congenital heart disease [20].

In addition, obstructive sleep apnea [21] and macroglossia [22] are generally found due to hypodevelopment of the middle and lower facial areas.

Although described anteriorly by Jean Etienne Dominique Esquirol and Édouard Séguin, John Langdon Haydon Down (1828-1896) described in details the syndrome named after him in 1866 [23] (figure 7).

Down was a British doctor from Torpoint, the son of a grocer with a pharmacy [24]. He was the medical superintendent of the Earlswood Asylum in Surrey. Considered as a liberal, and a man with charming

manners, numerous of his dissertations are relevant to mental health

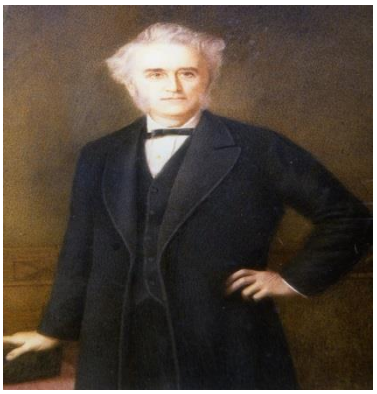


Figure 7. John Langdon Haydon Down.

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Paget´s disease of bone

Sir James Paget (1814-1899) was an English surgeon and pathologist from Norfolk (figure 8).

Figure 8. James Paget

He is considered to be one of the founders of scientific medical pathology, explaining and naming distinct conditions: Paget´s disease of bone (1877) [25], extramammary Paget´s disease, Paget-Schroetter disease, and Paget´s abscess. Paget´s disease of bone is a criterion for dysregulated bone remodelling, a painful scenario resulting from periosteal irritation [26]. Sir James Paget graduated in 1836 and became a member of the Royal College of Surgeons that same year. He later became a curator of the College of Surgeons Anatomy Museum. He had a very successful private practice, was an excellent speaker, and was elected a Fellow of the Royal Society, later Vice-President and President. Sir Paget was a nobleman, 1st baronet, and a friend of Charles Darwin and Thomas Henry Huxley. James Paget University Hospital in Norfolk, England bears his name in homage.

Marfan syndrome

Antoine Bernard-Jean Marfan (pronunciation: maʁfɑ̃, 1858-1942) was a French pediatrician from Castelnaudary (figure 9).

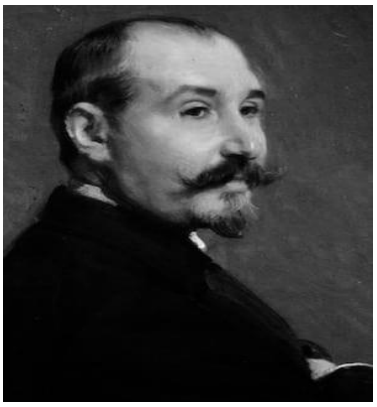

Figure 9. Antoine Bernard-Jean Marfan.

As a son of a medical practitioner, his father discouraged him from studying medicine. Nominated as a professor at the University of Paris, much of his career was at the Hôpital des Enfants Malades. In 1896, described Marfan syndrome [27], presenting a case of a girl to the *Société Médicale des Hôpitaux de Paris*. It is an autosomal dominant disorder, characterized by cardiovascular disease, eye disease, and skeletal features such as excessive height, arachnodactyly, and *pectus excavatum* [28]. In addition, some athletes who practice sports where tall stature is critical, such as basketball and volleyball, have Marfan syndrome [29].

Treacher Collins syndrome

Edward Treacher Collins (1862-1932) was a British surgeon and ophthalmologist from London (figure 10).

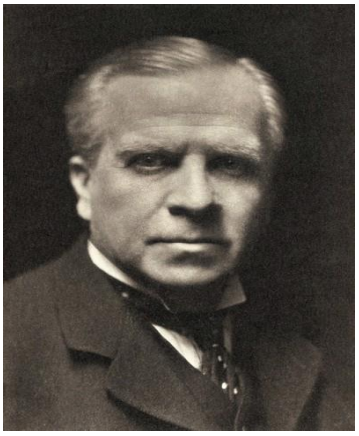

Figure 10.Edward Treacher Collins

As the son of a doctor, Dr. William Job Collins and Mary Ann Francisca Treacher, his name must be graphed without a hyphen.
He described Treacher Collins syndrome in 1900 at a meeting of the Ophthalmological Society, in London [30]. An autosomal dominant disease characterized by underdevelopment of the middle and lower face, malformations of the outer and middle ear with hearing loss in some cases, and tooth anomalies [31]. In his early career, he was asked to evaluate Persian Shah´s eldest son and the success of this Persian journey helped him greatly in his medical career [32]. A prominent figure in the organization of the British Ophthalmological Society and later President. Was considered as an outstanding rugby player, horse rider, and painter.

Apert syndrome

Eugène Charles Apert (1868-1940) was a French pediatrician from Paris, specialized in genetic disorders and congenital anomalies (figure 11).

Figure 11.Eugène Charles Apert

In 1906 he first described [33] a syndrome in children with craniosynostosis, midface hypoplasia, pseudomandibular prognathy, and syndactyly [34]. He was a pupil of Antoine Bernard-Jean Marfan and also served in World War I. As a founding member of French Society of Eugenics, he published a handbook on raising child that is still read by French mothers.

Crouzon syndrome

Louis Édouard Octave Crouzon (1874-1938) was a French neurologist from Paris, specialized in spinocerebellar ataxia [35] (figure 12).

Figure 12.**Louis Édouard Octave Crouzon.**

He was the first to present a syndrome in 1912 [36] initially known as craniofacial dysostosis, a mild form of many craniosynostosis syndromes. This syndrome is characterized by premature fusion of the cranial and facial sutures, increased intracranial pressure, exorbitism and midface hypoplasia [37]. During the First World War he was used in the rescue service. He was president of the *Société Neurologique de Paris*. Described as kind, generous, hardworking and patient.

Frey´s syndrome

Łucja Frey (1889-1942) was a Polish neurologist (figure 13).

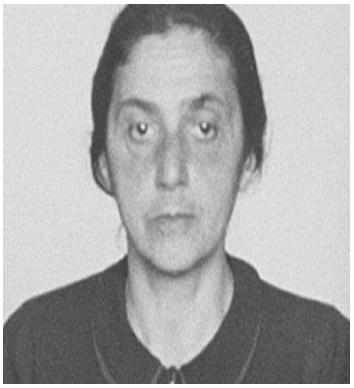

Figure 13.Łucja Frey, 1941.

She was born in the Polish city of Lwów, now Lviv in Ukraine. Frey´s syndrome is a neurological disease due to a parotid gland or auriculotemporal nerve damages that causes focal hyperhidrosis, with redness and sweating on the preauricular area. This syndrome was already described by Baillarger and Dupuy, but in 1932 it was Łucja Frey [38] who described the pathomechanism, clarified it and applied the term "auriculotemporal syndrome" [39]. Frey was one of the first female neurologists in Europe, noted as calm, humble, hard-working and methodical. She was of Jewish origin, was unfortunately deported to an extermination camp during World War II and possibly died in 1942.

Pierre Robin sequence

Pierre Robin (pronunciation: pjɛʁ ʁɔbɛ̃, 1867-1950) was a French dental surgeon from Paris. Professor at the French School of Stomatology and Editor-in-Chief of the *Revue de Stomatology*. He described the Pierre Robin´s sequence in 1923 [40], characterized by a U-shaped cleft palate, micrognathia and glossoptosis. A *Rue Pierre Robin* (street) in Lyon, France was named after him. Little is known about his private life.

Behçet´s disease

Behçet´s disease is a multisystem inflammatory disease that is well-known in Middle East and Asia [41]. It was originally described in 1937 [42] by the Turkish dermatologist Hulusi Behçet (pronunciation: bɛˈʧɛt, 1889-1948) from Istanbul. Son of a respected businessman, trained in Syria and Turkey, fluent in Turkish, French, Latin and German. He served in World War I and later worked in Hungary and Germany, with more than 200 published scientific papers. He was honored by the Turkish government with a depicted national postcard stamp (figure 14).

Figure 14. Hulusi Behçet

Eagle syndrome

The styloid process of the temporal bone leghtening or stylohyoid ligament calcification (figure 15) is referred to as Eagle syndrome [43] and was first cited in 1937 by American otorhinolaryngologist Watt Weems Eagle (1898 - 1980), from Statesville [44] (figure 16).

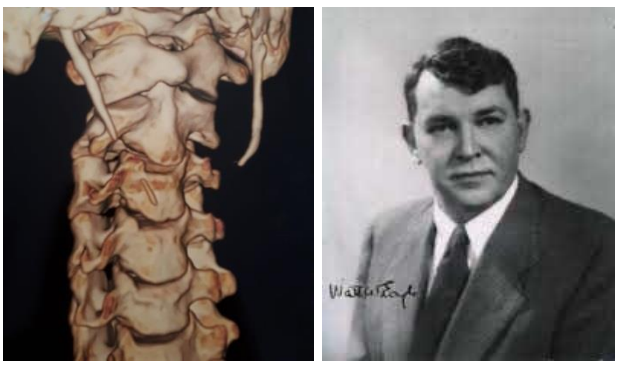

Figure 15. A calcified styloid process in a tomographic view
Fig.16 Watt Weems Eagle

It causes pain upon turning the neck or sticking the tongue. He completed his otolaryngology residence at Johns Hopkins and was First Chief, spending most of his career at Duke Medical Center [45].

Sjögren´s syndrome

Henrik Samuel Conrad Sjögren (pronunciation: ʃoʊgrɛn, 1899-1986) was a Swedish ophthalmologist from Köping (figure 17) who in his doctoral thesis in 1938 [46], defined the syndrome named after him.

Figure 17.Henrik Samuel Conrad Sjögren.

An autoimmune disease characterized by moisture producing, dry skin, chronic cough, numbness of the limbs and a high risk of lymphoma [47]. He has been a visiting lecturer at the University of Gotheburg, an honorary member of the Australian Ophthalmological Society, the American Rheumatism Organization, and the Royal College of Physicians and Surgeons of Glasgow.

He should not be confused with Karl Gustaf Torsten Sjögren, a Swedish psychiatrist and geneticist who named both Sjögren-Larsson syndrome and Marinesco-Sjögren syndrome.

Goldenhar syndrome

First described by Von Arlt in 1845, it was only recognized as an entity in 1952 [48] when the Belgian-American ophthalmologist, Maurice Goldenhar (1924-2001), described the syndrome that bears his name [49]. Also known as "first arch syndrome" or oculo-auriculo-vertebral dysplasia, the yet more widely used and conveniently recognizable name is Goldenhar's syndrome. The syndrome is characterized explicitly by peribulbar and/or labial dermoids, atrial appendages, and atrial fistulas with a blind bottom located in the pre-tragus region, microtia, and vertebral anomalies [50,51]. Maurice emigrated from Belgium to the United States of America in 1940 due to World War II. However, his medical dissertation was performed at the University of Geneva what produces some confusion leading to some person believe he was born in Switzerland.

Gorlin-Goltz syndrome

Basal cell nevus syndrome, basal cell carcinoma nevus syndrome, Gorlin syndrome, Gorlin-Goltz syndrome or nevoid basal cell carcinoma, is a rare autosomal dominant disease [52], described in 1960 [53]. Signs and symptoms include multiple basal cell carcinomas , odontogenic

keratocysts, and skeletal diseases such as hypertelorism and mandibular prognathism [54]. This is shown in Figure 18.

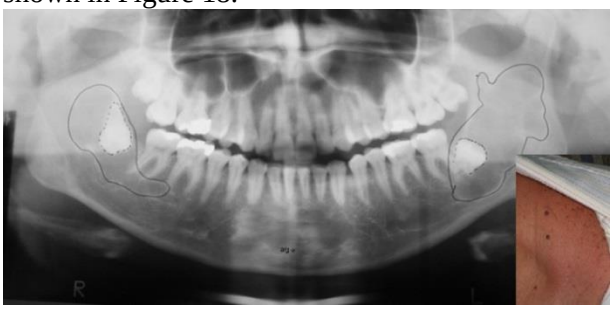

Figure 18. Panoramic radiography with multiple keratocysts. Insert: Presence of multiple nevus in the same patient, diagnosed as a Gorlin-Goltz syndrome.

Robert James Gorlin (1923-2006) was an American oral pathologist and geneticist from Hudson, New York (figure 19).

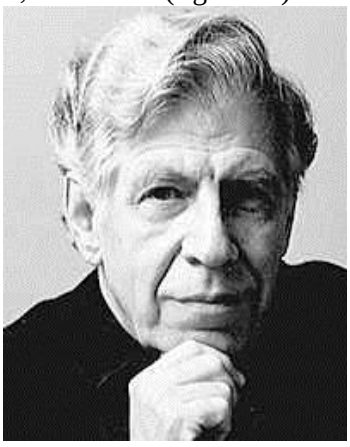

Figure 19.Robert James Gorlin.

Served in the US Army during World War II and also completed graduation and doctorate in dentistry, master´s degree in chemistry. Chairman of Oral Pathology at the University of Minnesota, has published more than 400 papers. He has served on the editorial board of impressive oral pathology journals and founding member of the American Board of Medical Genetics.

Robert William Goltz (1923-2014) was an American dermatologist from Minnesota (figure 20).

Figure 20.Robert William Goltz

He was the University of Colorado´s the first professor of dermatology in 1965, and retired as chairman of the University of Minnesota Chairman in 1985. A pioneer in dermatopathology.

Williams syndrome

John Cyprian Phipps Williams (1922-?) is a New Zealand cardiologist (figure 21), best known for Williams syndrome, originally described in 1961 [55].

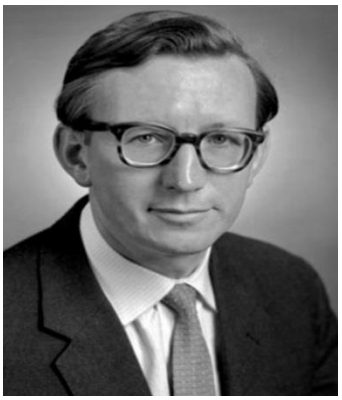

Figure 21. John Cyprian Phipps Williams.

Patients affected by Williams syndrome are undersized and have a unique face, with one constant facial expression of happiness [56]; this is why it is called "Happiness syndrome". In addition to heart and behavioral characteristics, Williams syndrome is associated with dental disorders (missing teeth, microdontia, changes in tooth shape) [57]. Dr Williams was working at Greenlane in 1964, when he received two offers from the Mayo Clinic, in the United States, and he never showed up. Dr. Williams has been reported missing and presumed dead by Interpol from 1978.

Pfeiffer syndrome

Rudolf Arthur Pfeiffer (1931-2012) was a German geneticist, from Saarbrücken who studied medicine in Saarbrücken, Vienna, Frankfurt, and Heidelberg. In 1964 [58] he defined the syndrome named after him, characterized by craniosynostosis, turribrachycephaly, proptosis, hypertelorism and midface hypoplasia [59]. With more than 500 scientific articles, he is one of the first authors to illustrate thalidomide embryopathy [60]. Founding member of the European Society for Genetic Counseling, was a music enthusiast and pianist.

Conclusion

The use of medical eponyms is secular. Although some authors recommend not use them, in certain scenarios such as syndromes or disorders, they can enable communication between experts and caregivers. In these succinct biographies there are histories of successful careers, and researchers whose lives were abruptly interrupted, but still managed to bold leave a legacy to mankind. The authors hope this article honors these pioneers whose shoulders we stand. This list of oral and craniomaxillofacial

syndromes and disorders is exhaustive and could be the focus of a future research project.

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