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EPONYMS RELATED TO GENETIC DISORDERS ASSOCIATED WITH GINGIVAL ENLARGEMENT; PART I

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Gingival enlargement is common among patients and can be caused by a variety of etiological factors. The most common reason is poor oral hygiene and high bacterial load that leads to gingival inflammation and enlargement. Other implicated factors include systemic drugs, such as Phenytoin, Nifedipine, Verapamil and Cyclosporine. Some enlargements could be associated with other conditions such as puberty, pregnancy or diabetes or be a symptom of a systemic disease (leukemia, Wegener's granulomatosis or sarcoidosis) [1].

There are also genetic disorders associated with gingival enlargement, which can be sorted into four groups, namely, Hereditary Gingival Fibromatosis (HGF), lysosomal storage disorders, vascular disorders and syndromes characterized by the presence of characteristic dental abnormalities .

Hereditary Gingival Fibromatosis (HGF), represents a heterogeneous group of disorders characterized by progressive enlargement of the gingiva. It manifests itself by an enlarged gingival tissue covering teeth to various extents. HGF may appear as an isolated entity i.e. as autosomal dominant Gingival Fibromatosis, which has little consequence apart from a cosmetic problem and occasional associations with hypertrichosis and/or epilepsy, or as part of a syndrome [2-4].

In Table I [5-16], we shed some lights on eponymous syndroms related to gingival fibromatosis.

Eponyms related to disorders associated with gingival fibromatosis	Remarks
Costello syndrome (CS) [5,6]	It is a distinctive rare multisystem disorder comprising a characteristic coarse facial appearance, intellectual disabilities, and tumor and papillomata predisposition. Heart abnormalities are also common. Although the diagnosis can be suspected clinically, confirmation requires identification of a heterozygous mutation in the proto-oncogene HRAS. Oral examination is important as CS patients develop gingival hyperplasia usually within the first years of life and is considered as a quite distinct feature that can also aid in its differential diagnosis from Noonan syndrome and Cardiofaciocutaneous syndrome that phenotypically overlap with CS. CS was discovered by Dr Jack Costello, (Fig. 1), a New Zealand Paediatrician in 1977. Dr Costello died in 2010.

Figure 1. Dr Jack Costello.

Table I. Eponyms related to disorders associated with gingival fibromatosis.

Figure 2. Dr Harold E. Cross.

Figure 3. Dr. Andreas Giedion.

Figure 4. Friedrich Daniel von Recklinghausen (1833-1910

Eponyms related to disorders associated with gingival fibromatosis	Remarks
Cowden syndrome [7,8]	Also known as, „Multiple hamartoma syndrome". It is a rare autosomal dominant inherited disorder characterized by multiple tumor-like growths called hamartomas and an increased risk of certain forms of cancer. Cowden syndrome is associated with loss-of-function mutations in PTEN, a tumor suppressor gene, leading to hyperactivity of the mTOR pathway. Cowden syndrome may cause oral papilloma rather than gingival swelling. Multiple traumatic fibromas, oral fibromas in tuberous sclerosis, Darier-White disease More Details, Heck's disease, lymphangioma, pyogenic granuloma, fibroepithelial polyps, lipoid proteinosis, oral florid papillomatosis, oral papillomas in Goltz syndrome, mucosal neuromas of multiple endocrine adenomatosis, acanthosis nigricans, pseudoepitheliomatous hyperplasia and squamous cell carcinoma should be considered in the differential diagnosis of oral papillomatous lesions Cowden syndrome was first described in 1963 by Lloyd & Dennis. They named the condition after the surname of their patient, Rachel Cowden.
Cross syndrome [3]	Also known s, Cross- McKusick- Breen syndrome or Kramer's syndrome. It is characterized by GE, nanophthalmos, microcornea, hypopigmentation, mental retardation and writhing movement of hands and legs . Named after, a USA ophthalmologist, working in University of Arizona, Harold E. Cross, (Fig. 2), who was born in 1937.
Jones syndrome [9,10]	It is an autosomal dominant disorder characterized by gingival fibromatosis with progressive sensorineural deafness. First reported by Jones et al, in 1977.
Murray-Puretic-Drescher syndrome [11]	This is another name for, Juvenile hyaline fibromatosis (JHF), which is a rare autosomal recessive disease characterized by papulonodular skin lesions, gingival hyperplasia, joint contractures, and bone lesions. JHF was for the first time described by Murray in 1873 and named by Drescher et al, in 1969.
Ramon syndrome [12]	This syndrome comprises the association of cherubism with gingival fibromatosis, epilepsy, mental retardation, stunted growth, and hypertrichosis.Named after an oral surgeon, Yochanan Ramon who and his colleagues reported the condition in 1967.
Rutherfurd syndrome [13]	It is a rare genetic disorder that is primarily characterised by the classical triad of gingival fibromatosis, delayed tooth eruption and corneal dystrophy. First reported, by Rutherfurd in 1931.
Schinzel-Giedion syndrome (SGS) [14]	It is a rare multiple congenital malformation syndrome defined by characteristic facial features, profound developmental delay, severe growth failure, and multiple congenital anomalies. Most individuals affected by SGS die in early childhood mainly because of progressive neurodegeneration and respiratory failure. However, a long-lived patient showed gingival hyperplasia that was progressive even after gingivectomy. The causative gene of SGS, SETBP1, was identified. SGS was first described in 1978 by an austrian geneticist, Dr. Albert Schinzel, born in 1944 and a Swiss radiologist, Dr. Andreas Giedion, (Fig. 3), born in 1925.

Table I. Eponyms related to disorders associated with gingival fibromatosis (continued).

Eponyms related to disorders associated with gingival fibromatosis	Remarks
von Recklinghausen syndrome [15]	This is another name for, Neurofibromatosis type 1 (NF1), which is a neurocutaneous disorder characterized by neural and cutaneous manifestations, as well as skeletal, oral and jaw abnormalities. This syndrome is named after, Friedrich Daniel von Recklinghausen (1833–1910), (Fig. 4), who was a German pathologist.
Zimmermann-Laband syndrome [16]	It is a very rare disorder characterized by gingival fibromatosis, abnormalities of soft cartilages of the nose and/or ears, hypoplastic or absent nails and terminal phalanges, joint hypermobility, hepatoslenomegaly, mild hirsutism and learning difficulties. Named after, Karl Wilhelm Zimmermann (1861-1935), (Fig. 5), who was a German anatomist and histologist, and Peter F. Laband, who was USA dentist born in 1900.  **Figure 5. Karl Wilhelm Zimmermann (1861-1935).**

Table I. Eponyms related to disorders associated with gingival fibromatosis (continued).

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