

Eponyms related to genetic disorders associated with gingival enlargement: Part II

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There are genetic disorders associated with gingival enlargement. In our part I, we reviewed the eponyms linked to Hereditary Gingival Fibromatosis (HGF) [1].

In this part II of this review, we are going to shed some light on eponyms linked to groups of genetic disorders which may feature gingival enlargement (Table I) [2-15].

Table 1: Eponyms related to genetic disorders associated with gingival enlargement

Eponyms related to genetic disorders associated with gingival enlargement	Remarks
Anderson – Fabry Disease [2,3]	Also known as Fabry disease, angiokeratoma corporis diffusum and alpha-galactosidase A deficiency. It is a rare X-linked lysosomal storage disease, which can cause a wide range of systemic symptoms. The disease is named for Johannes Fabry (1860-1930), (Fig. 1), who was a German dermatologist. And William Anderson (1842-1900), (Fig. 2), who was an English surgeon and dermatologist. In this disease the angiokeratomas may also involve the oral mucous membrane and the gingiva. Histologically, angiokeratoma of the gingiva shows ceramide inclusions not only in the connective tissue, but also in the oral epithelial cells
Goltz syndrome [4,5]	This is another name for, focal dermal hypoplasia. Also called Goltz-Gorlin syndrome. It is a rare syndrome and may result in multisystem disorders. Robert William Goltz, (Fig. 3), is an American dermatologist, born 1923. Robert James Gorlin (1923-2006), (Fig. 4), was an American oral pathologist and geneticist. Partial anodontia is the characteristic dental feature. Other oral manifestations include lip papillomas, gingival enlargement and hypoplastic teeth. Histopathologic features showed deposits of fat cells or adipose tissue in the dermis
Hunter syndrome [6]	Also known as Mucopolysaccharidosis II. It is a rare, X-linked disease caused by a deficiency of the lysosomal enzyme iduronate-2-sulfatase, which catalyses a step in the catabolism of glycosaminoglycans. The glycosaminoglycans accumulate within tissues affecting multiple organs and physiologic systems. The clinical manifestations include neurologic involvement, severe airways obstruction, skeletal deformities and cardiomyopathy. Hunter syndrome presents the same oral manifestations as Hurler syndrome. Hunter syndrome is named after physician Charles A. Hunter (1873-1955), who first described it in 1917. Born in Scotland, Hunter emigrated to Canada and had a medical practice in Winnipeg, Manitoba
Hurler syndrome [7-9]	This is another name for Mucopolysaccharidosis type I (MPS I). It is one of the most frequent lysosomal storage diseases. It has a high morbidity and mortality, causing in many cases severe neurological and somatic damage in the first years of life. MPS I involves a broad spectrum of clinical symptoms and can be divided into three clinical subtypes: Hurler, Hurler/Scheie, and Scheie syndromes. Scheie syndrome is considered a less severe version of Hurler syndrome. Gingival hyperplasia is a common feature in Hurler syndrome. Other intraoral features include macroglossia, spaced hypoplastic peg-shaped teeth with retarded eruption. Gertrud Hurler (1889-1965), (Fig. 5), was a German Pediatrician and a Medical practitioner. Harold Glendon Scheie (1909-1990), (Fig. 6), was an American Ophthalmologist
Klippel-Trenaunay Syndrome (KTS) [10]	It is an uncommon mesodermal phakomatosis characterized by a triad of cutaneous and visceral hemangiomas, venous varicosities and soft tissue or bone hypertrophy. It was first described, in 1900, by French physicians Maurice Klippel (1858-1942) and Paul Trénaunay. Frederick Parkes Weber (1863-1962), (Fig. 7), who was an English dermatologist described cases in 1907 and 1918 that were similar but not identical to those described by Klippel and Trénaunay. KTS may be associated with many oral abnormalities such as gingival enlargement, gingival capillary hemangiomas, gingival fibroma, gingival fibromatosis, gingival hyperplasia, high arched palate, unilateral hypertrophy, or increase in size of periodontal tissues, tongue capillary hemangiomas and unilateral macroglossia

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Table 1: continued...

Eponyms related to genetic disorders associated with gingival enlargement	Remarks
Maroteaux-Lamy Syndrome [11]	This is another name for Mucopolysaccharidosis VI (MPS VI). It is an inheritable, clinically heterogeneous lysosomal storage disorder that develops due to a deficiency in the arylsulfatase B (ASB) enzyme. This deficiency impairs the stepwise degradation of glycosaminoglycans (GAGs) resulting in the accumulation of partially degraded GAGs in tissues and organs throughout the body. The oral findings include short or stubby, malformed, peg-shaped, poorly formed and calcified teeth with delayed eruption and gingival hyperplasia. It is named after Pierre Maroteaux (1926-), (Fig. 8), and his mentor Maurice Emil Joseph Lamy (1895-1975), both French physicians
Menkes Kinky hair Disease [12]	It is a very rare X-chromosome disorder affecting copper metabolism. Menkes et al. in 1962, first described the disease in a family of English-Irish descent living in New York, who presented by early retardation in growth, peculiar hair, and focal cerebral and cerebellar degeneration. The disease leads to psychomotor retardation, pili torti, pallor, seizures and death in infancy or early childhood. The oral findings include delayed dentition and gingival hyperplasia. The disease is named after John Hans Menkes (1928-2008) (Fig. 9). Menkes, was born in Vienna. In 1939, following the German annexation of Austria, the family fled Austria and immigrated to the USA via Ireland. Menkes was then eleven years old. Menkes, son and grandson of physicians, followed the family tradition and studied medicine, despite his wish to become a journalist. Menkes became head of pediatric neurology at the Johns Hopkins Hospital and later at the University of California, Los Angeles (UCLA)
Niemann - Pick disease [13]	This disease is a form of sphingolipidosis, which in turn, are included in the larger family of lysosomal storage diseases (LSDs). Oral findings include thick lips, macroglossia and widely spaced teeth. Gingival enlargement is not considered a constant feature. Albert Niemann (1880-1921), who was a German physician published the first description of what is now known as Niemann-Pick disease, type A, in 1914. Ludwig Pick (1868-1944), (Fig. 10), who was a German pathologist described the pathology of the disease in a series of papers in the 1930s
Sly syndrome [14]	This is another name for Mucopolysaccharidosis Type VII. It is an autosomal recessive lysosomal storage disease characterized by a deficiency of the enzyme β -glucuronidase, a lysosomal enzyme. Sly syndrome belongs to a group of disorders known as mucopolysaccharidoses, which are lysosomal storage diseases. The oral features include mainly thickening of the alveolar ridges and rarely gingival hyperplasia. It was named after its discoverer William S. Sly (1932-), (Fig. 11), an American Biochemist, in 1969 who has spent nearly his entire academic career at Saint Louis University
Sturge Weber syndrome [10]	It is a rare sporadic condition of mesodermal phakomatosis, characterized by purple-colored flat cutaneous cranial (face) hemangiomas (most commonly along the trigeminal nerve), glaucoma and vascular lesions in the ipsilateral brain and meninges. One of its features is unilateral vascular hyperplasia of oral mucosa and gingiva. It is named for William Allen Sturge (1850-1919), (Fig. 12), who was an English physician and Frederick Parkes Weber (1863-1962), (Fig. 7), who was an English dermatologist
Wilson syndrome [15]	Wilson disease (WD) is a genetic disorder with copper metabolism disturbances leading to copper accumulation in many organs with their secondary damage. It is caused by mutation in the ATP7B gene on chromosome 13, which encodes ATP-ase 7B involved in copper transport. It is characterized by multiple small red papules of the lips, gingival enlargement, early onset periodontitis, and repeated oral candidiasis. Enamel hypoplasia is the characteristic dental feature. Wilson's disease is named after Samuel Alexander Kinnier Wilson (1878-1937) (Fig. 13), the British neurologist who first described the condition in 1912. Kayser-Fleischer rings (KF rings) are dark rings that appear to encircle the iris of the eye. They are due to copper deposition in Descemet's membrane as a result of this disease. They are named after Dr. Bernhard Kayser (1869-1954) and Dr. Bruno Fleischer (1874-1965), (Fig. 14), the German doctors who first described them in 1902 and 1903. Initially thought to be due to the accumulation of silver, they were first demonstrated to contain copper in 1934



Figure 1: Johannes Fabry (1860-1930)

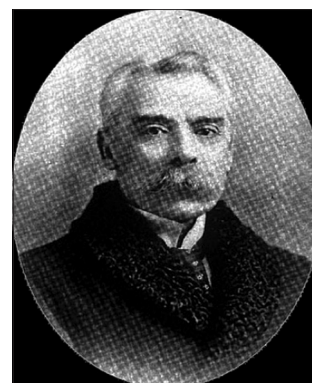


Figure 2: William Anderson (1842-1900)



Figure 3: Robert William Goltz

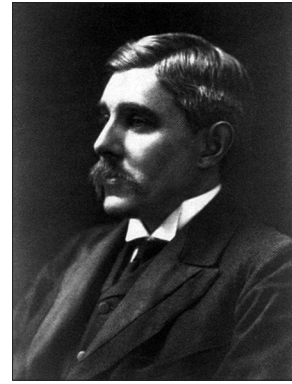


Figure 7: Frederick Parkes Weber (1863-1962)



Figure 4: Robert James Gorlin (1923-2006)



Figure 8: Pierre Maroteaux (1926-)



Figure 5: Gertrud Hurler (1889–1965)

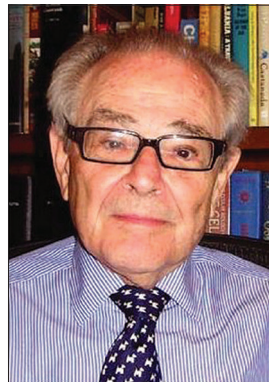


Figure 9: John Hans Menkes (1928-2008). Reprinted with permission from *Neurology Today*, an official publication of the American Academy of Neurology



Figure 6: Harold Glendon Scheie (1909-1990)

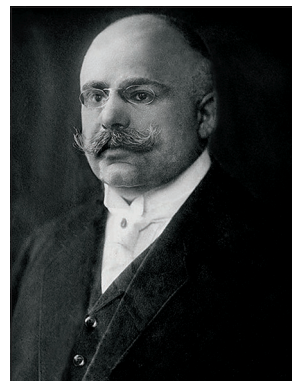


Figure 10: Ludwig Pick (1868-1944)



Figure 11: William S. Sly



Figure 13: Samuel Alexander Kinnier Wilson (1878-1937)



Figure 12: William Allen Sturge (1850-1919)



Figure 14: Bruno Fleischer (1874-1965)

These groups include lysosomal storage disorders, vascular disorders and syndromes characterized by the presence of characteristic dental abnormalities.

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