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SPECIAL REVIEW

Marathon of eponyms: 7 Gorlin–Goltz syndrome (Naevoid basal-cell carcinoma syndrome)

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The use of eponyms has long been contentious, but many remain in common use, as discussed elsewhere (Editorial: Oral Diseases. 2009: 15; 185). The use of eponyms in diseases of the head and neck is found mainly in specialties dealing with medically compromised individuals (paediatric dentistry, special care dentistry, oral and maxillofacial medicine, oral and maxillofacial pathology, oral and maxillofacial radiology and oral and maxillofacial surgery) and particularly by hospital-centred practitioners. This series has selected some of the more recognized relevant eponymous conditions and presents them alphabetically. The information is based largely on data available from MEDLINE and a number of internet websites as noted below: the authors would welcome any corrections. This document summarizes data about Gorlin–Goltz syndrome.

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Also known as

Basal cell naevus (carcinoma) syndrome
Epithélioma naevique multiple
Fifth phakomatosis
Gorlin syndrome
Hereditary cutaneo-mandibular polyoncosis
Hermans–Grosfeld–Spaas–Valk syndrome
Multiple basal-cell carcinoma syndrome
Multiple basal-cell naevi syndrome
Multiple hereditary cutaneomandibular polyoncosis
Multiple naevoid basal-cell carcinoma syndrome
Naevus epitheliomatodes multiplex
Naevoid basal-cell carcinoma syndrome
Naevoid basal-cell epithelioma-jaw cyst-bifid rib syndrome
Ward syndrome II

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The condition

The naevoid basal-cell carcinoma syndrome (NBCCS) is an autosomal dominant inheritable condition due to mutations in the Patched (*PTCH*) gene found on chromosome 9q, which affects the skin, jaws and other bones, nervous system, eyes and endocrine system, with high penetrance and variable expressivity.

Skin lesions appear in childhood or adolescence. Multiple naevoid basal-cell carcinomas over the nose, eyelids, cheeks and elsewhere are often an early sign. There may also be pitting of the palms or soles, and basal cell carcinomas can originate in these areas. Multiple jaw keratocystic odontogenic tumours (KCOTs) are seen in 75–90% of patients with NBCCS (these are the most prevalent abnormality), and anomalies of the vertebrae and ribs and many other bone abnormalities may be associated. Calcification of the falx cerebri is pathognomonic.There is frontal and temporoparietal bossing, a broad nasal root, prominent supra-orbital ridges and a degree of mandibular prognathism. Short fourth metacarpals are common.

There can be learning impairment in about 5% of the patients or cerebral tumours, particularly medulloblastomas, in about 5% of them. Pseudohypoparathyroidism has been described in some patients and there is also a slightly greater incidence of diabetes mellitus. Cardiac lesions may be present and cerebral tumours can be fatal. There is an associated increased incidence of ovarian fibromas.

Main differential diagnoses include Bazex syndrome (a rare acral psoriasiform dermatosis associated with internal malignancy, most frequently carcinoma of the upper aerodigestive tract), trichoepithelioma papulosum multiplex (an autosomal dominant disorder of multiple trichoepitheliomas) and Torre syndrome (Muir–Torre syndrome: association of skin sebaceous tumours with internal malignancy). Diagnosis of NBCCS is suggested by major criteria – positive family history; more than one basal cell carcinoma; KCOTs (first sign in 75% of the patients); palmar or plantar pits and calcified falx cerebri. Minor criteria include congenital skeletal anomalies: bifid, fused, splayed or missing ribs; or bifid, wedged or fused vertebrae; occipitofrontal

Table 1 Eponyms associated with Gorlin

Eponym	*Main features*
Goltz–Gorlin syndrome	Complex ectodermal and mesodermal dysplasia with bone involvement
Gorlin cyst	Benign odontogenic cyst histologically characterized by ghost bodies, assumed to be keratinized cells and an accentuated basal cell layer
Gorlin sign	Ability to touch the tip of the nose with the tongue in Ehlers–Danlos syndrome
Gorlin syndrome	Ocular, craniofacial, dental, hand and foot malformations and mildly delayed mental development
Gorlin syndrome II	Lentigenes, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonary stenosis, abnormal genitalia, dwarfism and sensorineural deafness
Gorlin syndrome III	Dwarfism, dislocated lenses, glaucoma and learning impairment
Gorlin–Chaudry–Moss syndrome	Craniosynostosis, midface hypoplasia, hypertrichosis and anomalies of heart, eyes, teeth and genitalia
Gorlin–Cohen syndrome	Tooth, bone and connective tissue abnormalities
Gorlin–Goltz syndrome	Basal cell naevi, jaw cysts, cranial enlargement and skeletal malformations
Gorlin–Holt syndrome	Tooth, bone and connective tissue abnormalities with facial asymmetry, pronounced supra-orbital ridges, wide nasal bridge, mandibular hypoplasia and small pointed chin
Gorlin–Pindborg syndrome	Orofacial, genital and musculoskeletal abnormalities, learning impairment, lower lip fistulas and creased skin on the back of the legs
Gorlin–Psaume syndrome	Orofacial, digital defects, CNS malformations, microcephaly, malformed eye canthi, defective alar naesi, webbed fingers, tremor, alopecia, lip and palate defects
Gorlin–Sedano syndrome	Short metacarpals and metatarsals, short terminal thumbs, short straight clavicles, and multiple impacted teeth
Gorlin–Vickers syndrome	Multiple mucosal neuromas, medullary carcinoma of the thyroid gland and marfanoid habitus

circumference over the 97th percentile, with frontal bossing; cardiac or ovarian fibromas; medulloblastoma; lymphomesenteric cysts and congenital malformations such as cleft lip and/or palate, polydactyly and congenital ocular anomalies (cataract, microphthalmos and coloboma).

Background to the eponym

Cases were described by Adolf Jarisch (1850–1902) in 1894, Moriz Kaposi (1837–1902) in 1872, Nomland in 1932 and Binkley and Johnson in 1951. The same syndrome was reported in 1960 by both Gorlin and Goltz, and by Ward, an Australian physician (Ward syndrome).

The main persons

Robert (Bob) James Gorlin was born on 11 January 1923, in Hudson, New York, USA.

He graduated at Columbia College, served in the US Army during World War II and then studied at Washington University School of Dentistry, graduating in 1947. He obtained an M.S. in Chemistry at the State University of Iowa. After a number of academic posts, Gorlin in 1956 moved to the University of Minnesota School of Dentistry, where he became professor and chairman of oral pathology in 1958 and Regents Professor in 1979. At the University of Minnesota, he served as professor of pathology, dermatology, paediatrics, obstetrics, gynaecology and otolaryngology.

Gorlin's major contribution was the delineation of craniofacial syndromes. His monograph Syndromes of the Head and Neck is the definitive work in the field. He is associated with a number of eponyms (Table 1).

Gorlin was one of the founders and a diplomate of the American Board of Medical Genetics, Clinical Genetics; a senior fellow of the Institute of Medicine at the National Academy of Sciences; served as editor of the oral pathology section of Oral Surgery, Oral Medicine and Oral Pathology and was on the editorial board of the American Journal of Medical Genetics and Dysmorphology and Clinical Genetics. He died on August 29, 2006, in Minneapolis, Minnesota.

Robert William Goltz, born in USA, studied Medicine at the University of Minnesota and in 1965 became the first professor of dermatology at the University of Colorado, Denver. In 1970 he returned to the University of Minnesota to head dermatology until he retired in 1985.

Associated persons

Robert William Goltz
Robert James Gorlin
William Herbert Ward

Source internet sites (accessed 21 February 2009) and further reading

Gorlin RJ, Goltz RW (1960). Multiple nevoid basal-cell epithelioma, jaw cysts and bifid rib. A syndrome. *N Engl J Med* **262:** 908–912.

Lo Muzio L (2008). Nevoid basal cell carcinoma syndrome (Gorlin syndrome). *Orphanet J Rare Dis* **3:** 32.

Scully C, Langdon J, Evans J (2009). Editorial. *Oral Dis* **15:** 185–186.

Ward WH (1960). Nevoid basal cell carcinoma associated with a dyskeratosis of the palms and soles. A new entity. *Aust J Dermatol* **5:** 204–207.

http://www.whonamedit.com, http://rarediseases.about.com/
http://medcosmos.blogspot.com/2008/09/1000-eponyms-in-medicine.html
http://insidesurgery.com/index.php?itemid = 264
http://en.wikipedia.org/wiki/List_of_eponyms