

HISTORY OF DERMATOLOGY SOCIETY NEWSLETTER
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Genodermatoses Among the Ancients: Basal Cell Nevus Syndrome in Egyptian Dynastic Brothers

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The concept of heritable traits and disease is by no means a new one. Long before Gregor Mendel, evidence suggests that selective breeding was used as far back as prehistoric times to improve crops and animal livestock traits. Genetics, from the Ancient Greek, γένεσις *genesis* or origin, is a discipline of biology and medicine, examining the heritable factors at play within and around us.

Basal cell nevus syndrome (BCNS), also known as Gorlin-Goltz syndrome, is a rare autosomal-dominant condition with complete penetrance and variable expressivity. It is the result in a defect in the expression of the gene *PTCH*. The disease was first reported in the literature by Jarisch and White in 1894. The spectrum of disease associated with this syndrome was described in detail by Gorlin in 1960¹; however, an archeologic finding documents this disease in the paleorecord more than 3000 years ago.

EGYPTIAN SKELETONS

Two Egyptian skeletons of the Dynastic period were excavated with findings consistent with BCNS.² Supporting evidence includes odontogenic cysts, bifid ribs, incompletely fused sacral laminae, brachymetacarpalia, and occipital asymmetry² (Figure 1, Figure 2, Figure 3).

DIAGNOSIS

Diagnosis of BCNS is made in the presence of 2 major criteria or 1 major and 2 minor criteria.³ The major criteria consist of the following: (1) >2 basal cell carcinomas or 1 basal cell carcinoma in patients younger than 20 years; (2) odontogenic keratocysts of the jaw (proven by histologic analysis); (3) ≥3 palmar or plantar pits; (4) bilamellar calcification of the falx cerebri; (5) bifid, fused, or markedly splayed ribs; and (6) first-degree relative with BCNS. The minor criteria include the following: (1) macrocephaly; (2) congenital malformations, such as cleft lip



Figure 1. Bifid rib. ©The Trustee, The Wellcome Trust. Reproduced with permission. Satinoff MI, Wells C. Multiple basal cell naevus syndrome in ancient Egypt. *Med Hist.* 1969;13:294–297.

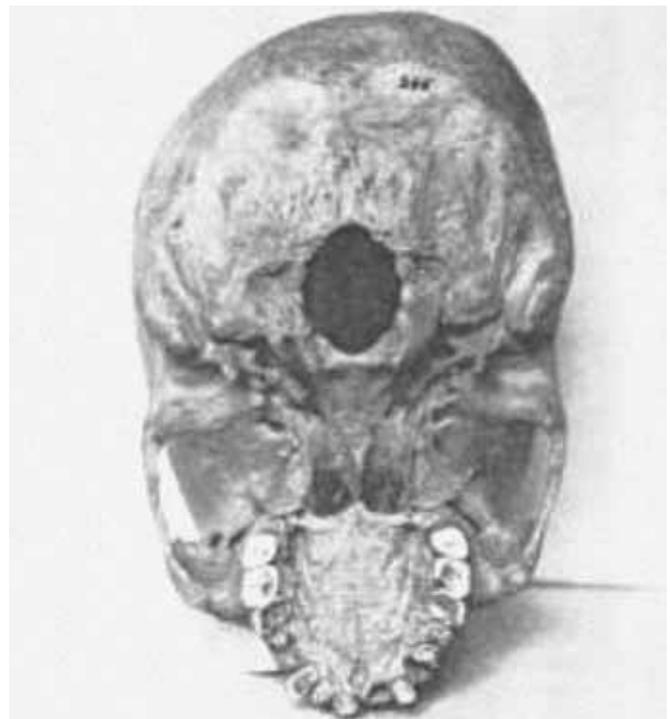


Figure 2. Asymmetry of the occipital bones. ©The Trustee, The Wellcome Trust. Reproduced with permission. Satinoff MI, Wells C. Multiple basal cell naevus syndrome in ancient Egypt. *Med Hist.* 1969;13:294–297.

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Figure 3. Brachymetacarpalia of the fourth digit. ©The Trustee, The Wellcome Trust. Reproduced with permission. Satinoff MI, Wells C. Multiple basal cell naevus syndrome in ancient Egypt. *Med Hist.* 1969;13:294–297.

sella turcica, vertebral anomalies, modeling defects of the hands and feet, or flame-shaped lucencies of the hands and the feet; and (5) ovarian fibroma or medulloblastoma. Typically, the condition is diagnosed with the presentation of multiple basal cell carcinomas or odontogenic keratocysts, both presenting in the second or third decade of life.^{4,5}

CONCLUSIONS

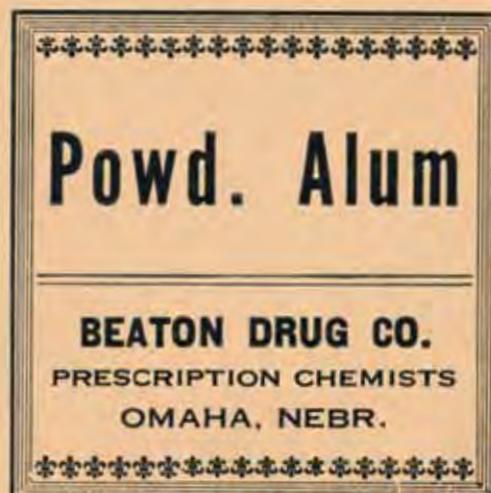
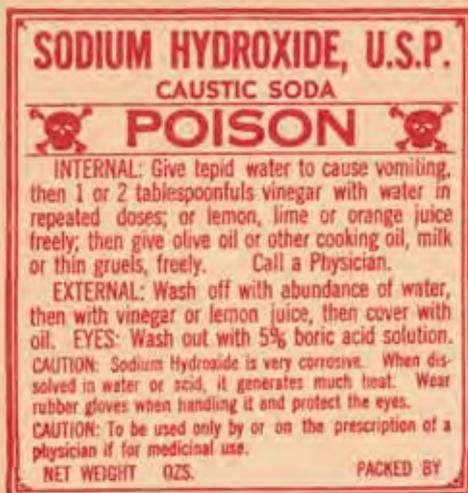
The cases presented support the diagnosis of BCNS with several major criteria met (nonhistologic evidence of odontogenic cyst, bifid rib, and affected family member) and are arguably the earliest documented cases of this genodermatosis, occurring 3000 years before Gorlin’s famous paper.

REFERENCES

- 1 Gorlin RJ, Goltz RW. Multiple nevoid basal cell epithelioma, jaw cysts and bifid rib: a syndrome. *N Engl J Med.* 1960;262:908–912.
- 2 Satinoff M, Wells C. The basal cell nevus syndrome in ancient Egypt. *Med Hist.* 1969;13:294–297.
- 3 Bitar GJ, Herman CK, Dahman MI, Hoard MA. Basal cell nevus syndrome: guidelines for early detection. *Am Fam Physician.* 2002;65:2501–2505.
- 4 Berg D. WebMD. Nevoid basal cell carcinoma syndrome. <http://emedicine.medscape.com/article/1101146-overview>. Updated March 12, 2009.
- 5 Kimonis VE, Goldstein AM, Pastakia B, et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. *Am J Med Genet.* 1997;69:299–308.

or palate, frontal bossing, coarse facies, and moderate or severe hypertelorism; (3) other skeletal abnormalities, such as Sprengel deformity, marked pectus deformity, and marked syndactyly of the digits; (4) radiologic abnormalities, such as bridging of the

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