

A Handy Update on Management of Basal Cell Nevus Syndrome

Revisiting an inherited autosomal dominant genodermatosis with emphasis on an important patient support group.

By Kristi Schmitt Burr and Robert T. Brodell, MD

Basal cell nevus syndrome (BCNS), also called Gorlin's Syndrome, is an autosomal dominant inherited genodermatosis involving multiple organ systems most commonly caused by mutation of the PTCH gene on chromosome 9q associated with multiple basal cell carcinomas. It occurs between one in 56,000 to one in 164,000 people. Any patient with five or more basal cell carcinomas, basal cell carcinoma before age 20, or a strong family history of multiple basal cell carcinomas may fit this phenotype; the condition should be considered in the differential diagnosis. Diagnosis of BCNS is based on clinical parameters. Since the PTCH gene is not present in 100 percent of patients with BCNS, its absence does not preclude the diagnosis.

Diagnosis

This spring we reviewed the diagnosis of BCNS, which is made by two major criteria or one major criterion and two minor criteria, which are recalled in Table 1. (For more details, see vol. 3, no. 4, p. 60. The mnemonic is slightly revised since original publication.)

Work-up of these patients could include biopsy of suspect skin lesions or jaw cysts. Furthermore, the clinician must obtain a complete history

BCNS Life Support Network
13463 Claridon Troy Road
Burton, Ohio 44021
bcns.org

and physical examination focused on extracutaneous manifestations. Skull x-ray, skeletal x-rays, dental Panorex film, MRI scan of head, and pelvic ultrasound are indicated. The patient shown in Figure 1 is 20-years-old and presented with multiple palmar pits 0.3-0.5mm in diameter. Identification of these pits—a major diagnostic criteria—led to accurate diagnosis of BCNS.

Management

Patient education is a critical component of management of patients with BCNS. In addition to careful follow-up and effective treatment of new BCCs, patients require instruction in sun avoidance and risk minimizing strategies. Patients often have questions about long-term prognosis and concerns about quality of life. Directing patients to a reliable support and education program, such as the BCNS Life Support Network (See note at left), is helpful. 

1. Gorlin RJ. Nevoid-basal-cell carcinoma syndrome. *Medicine (Baltimore)* 1987; 66:98-113.
2. Evans DG, Ladusans EJ, Rimmer S, Burnell LD, Thakker N, Frandon PA. Complications of naevoid basal cell carcinoma syndrome: Results of a population based study. *J Med Genet.* 1193; 30:460-4.
3. Amlashi SF, Riffaud L, Brassier G, Morandi X. Nevoid basal cell carcinoma syndrome: relation with desmoplastic medulloblastoma in infancy. A Population based review of the literature. *Cancer.* 2003; 98:618-24.
4. Gallani M, Leffell DJ, Bale AE. Evidence for a tumor suppressor gene on chromosome 9 in basal cell carcinomas of the skin. *Am J Hum Gen* 49 suppl:454. 1991.
5. Lo Muzio L, Nocini P, Buccì P, Pannone G, Consolo U, Procaccini M. Early Diagnosis of nevoid basal cell carcinoma syndrome. *J Am Dent Assoc* 1999.



Figure 1: Palmar pits 0.3-0.5mm in diameter present in a 20 year-old patient with BCNS.

Table 1: Criteria for BCNS

The condition is complex, but a simple mnemonic can help recall important criteria for diagnosis.

- B**asal cell carcinomas (multiple or early age onset)
- A**natomic changes (cleft lip, macrocephaly, vertebral fusion, bifid ribs)
- S**kin manifestation – palmar pits
- A**bnormal facies (hypertelorism, frontal bossing, macrocephaly)
- L**oss of bone in jaw (odontogenic keratocysts)
- C**orneal opacities and cataracts
- E**xpansile growth (ovarian fibroma and medulloblastoma)