

International Medical Association “Bulgaria” (IMAB)

# ***Journal of IMAB***

**Annual Proceeding  
(Scientific Papers)**

**2007**

**book 1**

ISSN: 1312-773X (Online)

*Publishers*  
*Peytchinski, Gospodin Iliev*  
*Pleven, Bulgaria*

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## A FAMILIAL CASE OF GORLIN-GOLTZ SYNDROME.

Ivelina Yordanova, Dimitar Gospodinov, Veselin Kirov\*, Verka Pavlova, Galja Radoslavova

Department of Dermatology and Venereology,

\*Department of Oncology,

Medical University – Pleven, Bulgaria

### SUMMARY:

Gorlin-Goltz syndrome (GGS) also known as Nevoid Basal Cell Carcinoma Syndrome is a rare autosomal-dominant disorder characterized mainly by the presence of multiple basal cell carcinomas (BCC), odontogenic keratocysts of the jaw and palmar pits. This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms. A case of familial Gorlin-Goltz syndrome with many of the common manifestations is reported. A 29-year-old woman and her 50-year-old mother with GGS are presented. The disease started respectively at the age of 18 and 22, with multiple basal cell carcinomas on the face and upper extremities. Because of multiple odontogenic keratocysts of the jaws they two have been treated surgically. Clinical, cranial CT, histological and dermoscopy images from both patients were obtained. Multiple BCC in the mother and the daughter were detected, nodular and superficial spreading, 10 - 30 mm in diameter. The daughter has many pits on her palms. Palmar pits have a characteristic dermoscopy with red globules inside the flesh-colored, slightly depressed lesions. The histological examinations revealed different histological variants of BCC. The X-rays examination showed two jaw cysts in the daughter, calcifications of the brain falx and bridges of the sella turcica in both patients. The BCC in the patients were treated with cryosurgery and surgical excisions with good results. The patients are followed up. In conclusion our case demonstrated multisystemic involvement of GGS. The combination of clinical, imaging and histological findings is helpful in identifying GGS patients. It is important to make an early diagnosis and a proper management in GGS, which may have cancer predisposition. The genealogical analysis is important for the determination of the genetic risk and the prognosis for the proband's relatives.

**Key words:** Gorlin-Goltz syndrome, basal cell carcinomas, jaw odontogenic keratocysts, palmar pits, dermoscopy, cryosurgery.

### INTRODUCTION:

Gorlin-Goltz syndrome also known as Nevoid Basal Cell Carcinoma Syndrome is a rare autosomal-dominant disorder characterized mainly by the presence of multiple

basal cell carcinomas, odontogenic keratocysts of the jaw and palmar pits. This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms (13). We present a case of familial Gorlin-Goltz Syndrome, characterized by multiple basal cell carcinomas, odontogenic keratocysts and skeletal manifestations.

### Case report:

A 29-year-old woman was admitted in our dermatological department with multiple basal cell carcinomas on the face and a 30 mm in diameter swelling in the right maxillary region. She had many pits on her palms. Her 50-year-old mother also had similar clinical symptoms. The disease started with small nodular basal cell carcinomas on the face and trunk when the daughter was 18 and the mother 22 years old. Because of recurrent skin carcinomas with or without pigment they both have been repeatedly treated surgically with transient result. They both have been operated on thrice because of odontogenic keratocysts of the jaws. Physical examination showed the patients were tall of stature – 180 and 185 cm respectively. Pathological changes affecting cardio-vascular, respiratory and the neural systems were not observed. Dermatological status in the daughter revealed multiple nodular basal cell carcinomas on the skin of her forehead, nose and temporal bone (fig. 1). She had multiple pigmented nevi on her trunk and many pits on her palms too. These pits had a characteristic dermoscopy with red globules mainly distributed inside the flesh-colored and slightly depressed lesions (fig.2, 3). Dermatological status in the mother showed multiple nodular basal cell carcinomas on the forehead, nose and chin 10 – 30 mm in diameter. Some of them were with eroded surfaces. She had two superficial spreading basal cell carcinomas in the infraorbital region on her left eye and her left arm (fig.4,5). Cranial CT revealed calcifications of the brain falx and bridges of the sella turcica in both patients (fig. 6 a, b). There were two jaw cysts in the daughter. Her right maxillary sinus was filled with a liquid formation 41/28 mm with a destructed frontal wall but without infiltration in the face tissues. A similar formation 27/25 mm without destruction and expansion was detected in the left maxillary sinus. There was a last molar retention (fig.7). The histological examinations revealed different