Skin transplant in patients with Gorlin-Goltz syndrome as a viable solution for covering skin defects

Authors: S. Arslanagic, R. Arslanagic
Clinical University Centre of Sarajevo

Basal cell naevus syndrome is a rare condition which is inherited as an autosomal dominant trait with complete penetrance and variable expressivity. The syndrome is found in all races. Morbidity and premature mortality are primarily related to the development of skin cancers and other tumors associated with the syndrome.

Men and women are affected equally.

The gene responsible has been localized to chromosome 9q22.1-q31. In the PTPN11 (patched) gene, tumor suppressor gene. The process of tumor development actually requires mutation in multiple growth control genes. Mutation in this gene may increase the risk of ovarian cancer. The disease has complete penetrance and variable expressivity (1,2). What causes these additional mutations to be acquired is unknown. Possible causes include chemical, physical, or biological environmental exposures (such as sunlight) or chance errors in cell replication.

The spectrum of disease associated with this syndrome was described in detail by Gorlin in 1960 (3).

Nevoid basal cell carcinoma syndrome is rare autosomal dominant disorder. A gene causing syndrome is identified, PTPN11 gene, tumor, is only known tumor suppressor gene whose mutations contribute to tumor development and to wide variety of malformations. The two prominent features of the syndrome are: 1. predisposition to basal cell carcinoma and tumors of other different organs, and 2. developmental defects manifested by congenital abnormalities of different organs and structures.

The hallmark of this disorder is the appearance of skin cancer, basal cell carcinoma, at or about puberty and age 35 years, but they may appear at a younger age. Skin manifestations include pits in the palm and soles, symmetrical palmar and/or plantar pits are seen in 67-82% of all people with basal cell nevus syndrome. When they do develop, they often do so early in life, being found in as many as 80% of patients younger than 10 years. Thus, they may be a helpful early criterion for the diagnosis basal cell naevus syndrome. More than 3 pits should be noted because the relevance of 1 or 2 pits may not be diagnostic.

Treatment for Gorlin’s syndrome

Patients with Gorlin syndrome often require ORL surgery to remove jaw cysts and facial basal cell carcinoma. One of the many other treatments available for basal cell carcinoma including cryotherapy, fluorouracil cream and timolol medication. They should not receive treatment with irradiation as this is liable to provoke the development of tumors. Some patients may require long-term treatment with irradiation such as isotretinoin or actretin.

Major criteria are:
- Multiple (>2) basal cell carcinomas at any age or one basal cell carcinoma less than 30 years or >10 basal cell naevi
- Histologically proven odontogenic keratocyst or polycystic bone cyst
- Palmar or plantar pits (2 or more)
- Ectopic calcification (increased fibrous tissue calcification of the maxilla or mandible)

Minor criteria are:
- Congenital skeletal defects: bifid, fused or missing rib
- Large head with occipito-frontal circumference > 97th percentile
- Frontal bossing
- Cardiac or ovarian fibroma
- Medulloblastoma
- Lymphoepithelial cysts
- Congenital malformation: cleft lip and/or palate

A diagnosis of Gorlin’s syndrome can be made if there are 2 major or 1 major and 1 minor criteria.

Case report

A patient, 49 year old woman with basal cell nevus syndrome. On oncogenic face of disorders, it is reported, with very extensive and very destructive multiple basal cell carcinoma, over 50 and ranging in size from 1 cm to 12 cm. The first signs of the syndrome occurred at the age of five. Most often carcinoma involved face, back, and chest but also extremities. Axillary and right side of nose is totally destructed. Silica...as was lower right eyelid. Silica...and partial lower left eyelid. There are very extensive, excoriated basal cell carcinoma on right side of face, right side of chest and back Silica... and also on skin left malar region. Besides, these skin changes, skeletal changes were very features of patient, especially deformities of thorax, scoliosis, and prominent ribs. Silica...pectus carinatum and prominent thoracolumbar spine. Also there were some anomalies: calcification of the face cerebellum and maxilla cyst. Surgeon excision and allograft transplantation was unsuccessful. Each graft is rejected. Besides surgery therapy, following therapy was administrated: keytruda, RTG therapy and local cytostatic therapy. Therapy with retinoid was discontinued after 20 days because multiple adverse effects. After hospital discharge, more was not answering the control.

Key words: Basal cell naevus syndrome, carcinoma basal cell, surgery therapy

References


Table 1. Summary of the main histopathological changes observed in the patient.

<table>
<thead>
<tr>
<th>Tumor Type</th>
<th>Description</th>
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<tbody>
<tr>
<td>Basal cell carcinoma</td>
<td>Multiple tumors ranging in size from 1 cm to 12 cm</td>
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<tr>
<td>Cystic lesions</td>
<td>Excoriated lesions involving face, chest, and back</td>
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<tr>
<td>Skeletal anomalies</td>
<td>Pectus carinatum and prominent thoracolumbar spine</td>
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