JSCR Journal of Surgical Case Reports

Journal of Surgical Case Reports, 2017;6, 1–3

doi: 10.1093/jscr/rjw224
Case Report

CASE REPORT

A multidisciplinary approach to the successful management of Gorlin syndrome

Ryan N. Mello, Zaki Khan, and Umar Choudry*

Division of Plastic and Reconstructive Surgery, Department of Surgery, University of Minnesota Medical Center, Minneapolis, MN, USA

*Corresponding address. Division of Plastic and Reconstructive Surgery, Department of Surgery, University of Minnesota Medical Center, Minneapolis, MN 55455, USA. Tel: +1-612-625-1933; Fax: +1-612-625-4441; E-mail: choud008@umn.edu

Abstract

Gorlin–Goltz syndrome (GGS) is a rare genetic syndrome with variable expressivity and autosomal dominant inheritance. The major features of GGS include numerous basal cell carcinomas (BCCs), keratocysts of the jaw, palmar/plantar pits and calcification of the falx cerebri. Authors report the case of a 51-year-old male with a 19-year history of GGS and multiple BCCs of the head and neck. He presented with a large ulcerating lesion on the right side of his face involving cutaneous, subcutaneous and muscular tissues of the temporal and orbital region. Additionally, magnetic resonance imaging revealed involvement of the right zygomatic bone, infratemporal fossa and mandible. This case is notable in that BCC invasion of the facial bones is rare. Extensive resection and reconstruction with a latissimus dorsi microvascular free muscle flap was performed. The success of this challenging case exemplifies the need for a multidisciplinary team that included dermatology, plastic surgery, oculoplastics and otolaryngology.

INTRODUCTION

Gorlin–Goltz Syndrome (GGS) or Nevoid Basal Cell Carcinoma Syndrome (NBCCS) is a syndrome with autosomal dominant inheritance. Approximately 70–80% of affected individuals inherit the condition from a parent, while the remaining 20–30% have a *de novo* mutation [1]. The prevalence is estimated to be as high as 1 in 31 000 [2], though this number varies greatly between studies [3]. A mutation in the PTCH1 [9q22.3] tumor suppressor gene has been identified as the genetic defect in GGS [4]. Diagnosis is commonly made using a combination of major and minor diagnostic criteria (Table 1) [1, 5, 6]. However, the most characteristic feature of GGS, and the focus of this report, is the many BCCs [3].

Given the numerous characteristics described by the major and minor criteria, it is evident that GGS poses a potential source of morbidity in affected individuals. The case presented in this report describes an individual with multiple BCCs including an invasive BCC of the temporal, pre-auricular, zygomatic and orbital areas. This is a unique case in that aggressive BCCs that invade the facial bones are rare [7]. Treatment of the temporal/orbital region required coordination among numerous medical specialties, culminating in excision of the BCC and then reconstruction with a latissimus dorsi microvascular free flap. This case demonstrates the need and effective implementation of a multidisciplinary medical team in the management of GGS.

CASE REPORT

A 51-year-old male, with a 19-year history of GGS, presented with a progressive skin lesion involving the right side of his face (Fig. 1). He had a history of multiple Moh's surgeries, excisions and a left facial reconstruction for numerous BCCs. Past medical history otherwise was unremarkable.

Received: October 24, 2016. **Accepted:** March 2, 2017

Table 1 Major and minor diagnostic criteria of GGS

Major criteria	Minor criteria
Calcification of falx cerebri	Childhood medulloblastoma
Jaw keratocyst	Lympho-mesenteric or pleural cysts
Palmar or plantar pits (two or more)	Macrocephaly
Multiple BCCs (BCC before age 30 or >5)	Cleft lip and/or palate
First-degree relative with GGS	Vertebral and/or rib anomalies
	Polydactyly
	Ovarian or cardiac fibromas
	Ocular anomalies

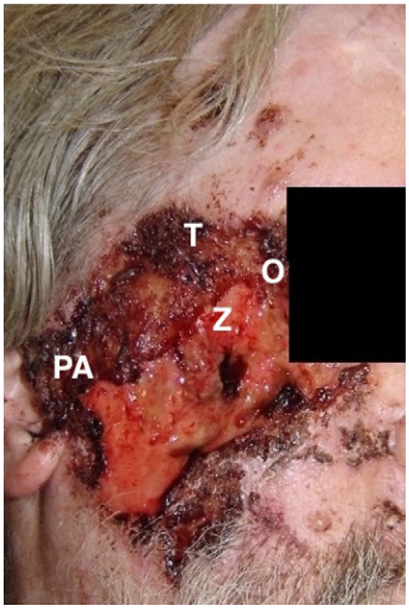

Figure 1: Preoperative image demonstrating large ulcerative lesion involving the temporal [T], pre-auricular [PA], zygomatic [Z] and orbital areas [O].

On physical examination, he had a large ulcerative lesion on the right side of the face, multiple small lesions on the scalp and upper torso, and multiple scars from previous surgeries. Visual acuity was normal in both eyes. There was purulent discharge and chemosis of the right eye. The lesion on the right side of the face involved the temporal, pre-auricular, zygomatic and orbital areas. Magnetic resonance imaging of the face showed involvement of right zygomatic bone, infratemporal fossa and mandible.

Numerous medical specialties were involved in the surgical management of the patient. The initial step surgically was resection of the lesion (Fig. 2). Oculoplastics performed resection of the bony orbital wall, peri-ocular muscles, and upper and lower eyelids of the right eye. Otolaryngology performed composite resection of skin, subcutaneous tissue, a portion of the right ear, a portion of the zygomatic bone and maxillary sinus. After these resections, plastic surgery completed a free flap reconstruction with the use of a right latissimus dorsi microvascular free muscle flap (Fig. 3). Six days after this first procedure, the patient underwent enucleation of the right eye (extensive resection had rendered the eye non-functional) with

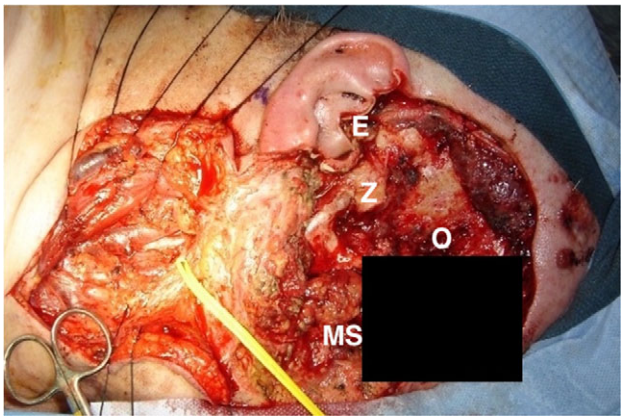

Figure 2: Intraoperative image after resection of skin, subcutaneous tissue, a portion of the right ear [E], upper and lower eyelids, peri-ocular muscles, bony orbital wall [O], a portion of the zygomatic bone [Z] and maxillary sinus [MS].

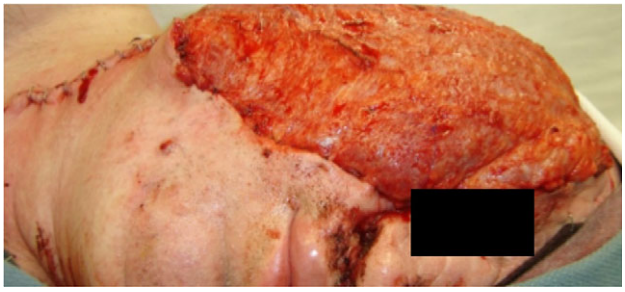

Figure 3: Intraoperative image after reconstruction of defect with a right latissimus dorsi microvascular free muscle flap.

re-elevation of the proximal portion of the free flap to cover the orbital cavity with muscle, along with skin grafting of the muscle flap. Final pathology of the specimen showed invasive BCC with focal squamous features, and negative surgical margins were confirmed. He was discharged 6 days after the second procedure.

In the months following discharge, four superficial lesions were removed via Moh's surgery, with primary closure and skin grafting where indicated. Regular follow-up appointments continue for the surveillance of new lesions with dermatology. The latissimus dorsi microvascular free muscle flap and skin graft remain viable and completely intact (Fig. 4).

DISCUSSION

The propensity to develop multiple BCCs is a potential source of morbidity in patients with GGS. BCCs in GGS patients range in number from a few to many thousands of lesions, with the face, back and chest being the most commonly involved sites [8]. The highest incidence of BCCs in these individuals is between puberty and age 35, with increased sun exposure believed to be a risk factor [9].

Surgical excision is often the treatment of choice for BCC, though several other modalities including cryotherapy and laser treatment may supplement surgery in the treatment of early lesions [1]. At the time of this publication, Vismodegib is indicated for metastatic BCC or recurrent BCC after surgery [10]. This chemotherapeutic option would have been ideal in this clinical scenario had it been available at the time. The patient was treated in 2010 and the FDA approved this drug two years

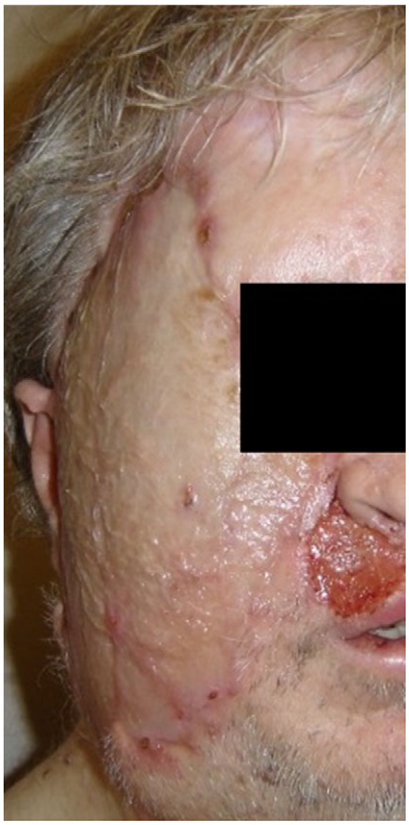

Figure 4: Three months post-operative image.

later in 2012 [10]. When surgery is deemed the first line of treatment, the goal should be to excise the entire lesion while sparing healthy tissue and when needed, use reconstructive surgery to maintain form and function. This challenge often requires the combined efforts of dermatology (including Moh's surgery), plastic surgery, otolaryngology, oculoplastics, dentistry and oral surgery, oncology and psychologists [3]. In addition to treating existing lesions, an important aspect in the management of GGS is prevention of new BCC lesions. Patients with GGS should be counselled regarding excessive sun exposure and the use of protective sunglasses, clothing and high factor sunscreens [1, 3].

GGS patients face considerable morbidity not only because of the manifestations of their syndrome, but also because of the treatment that may result in cosmetic and/or functional deficits. Clear communication should exist between the different physicians managing the different aspects of the patient's complex and challenging disease. A multidisciplinary approach is important to provide the best available care to the patient. This management starts with the proper diagnosis of the disease. The appropriate education of the patient and family regarding the lifelong management of the various aspects of this malady must be fulfilled by dermatology, genetics and various surgical specialties who will treat this disease entity. Specific treatment of the multiple BCCs and/or keratocysts the patients develop is another aspect of their treatment that often requires multispecialty expertise. Successful management of these complicated patients is definitely possible but requires early and sustained involvement of various experts, with clear communication and teamwork.

CONFLICT OF INTEREST STATEMENT

None declared.

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