Nevoid Basal Cell Carcinoma Syndrome: Report of 3 Cases

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Introduction

Nevoid basal cell carcinoma syndrome (NBCC), also commonly known as Gorlin-Goltz syndrome, is a rare autosomal dominant condition that presents with multiple neoplasms and developmental abnormalities. NBCC is a clinical diagnosis. We present 3 cases of NBCC with characteristic clinical features to create awareness of this syndrome to enable early diagnosis and treatment.

Case Report

Patient 1 is a 34-year-old man who was referred to us for multiple basal cell carcinoma (BCC) in 2013. He was diagnosed with medulloblastoma at age 3 and had his first BCC at age 12 which was surgically excised. Clinicopathological correlation supported Gorlin-Goltz syndrome as a diagnosis, but no further follow up was given. He is under dental follow up since the age of 20 for multiple odontogenic keratocysts which were surgically removed. In 2010, at age 29, he had a non-healing ulcer on his scalp and biopsy showed BCC which resolved with radiotherapy (Figure 1a)

Physical examination revealed frontal bossing, high-arched palate and numerous BCC on his head and upper trunk (Figure 1a-d). His BCCs were removed via curettage and electrodessication and he is on acitretin for chemoprophylaxis. Radioimaging shows odontogenic keratocysts in his jaw and calcified falx cerebri (Figure 1e-1g).

Patient 2 is a 48-year-old man with numerous recurrent “moles” treated in beauty salons since his mid 20’s. He has multiple odontogenic keratocysts since 18-years-old and was referred to us by dental team for multiple BCC on his face and scalp in 2014. At presentation, the largest BCC with central ulceration (3 cm x2cm) was removed by plastic surgery team. Other smaller BCCs were removed via curettage and electrodessication. He has hypertelorism, palmar pitting and skull x-rays shows falx cerebri calcification (Figure 2a-2c). He is on acitretin for chemoprophylaxis.

Patient 3 is a 47 year old man who had only 3 BCC excised since 41 year old. He has been under the follow up of dental team for odontogenic keratocysts since CT scan. CT scan noted to have calcification of falx cerebri in view of his minimal BCC, no chemoprophylaxis was given but he was advised for sun protection.

Discussion:

NBCC is caused by mutation of the sonic hedgehog pathway, leading to tumorigenesis and developmental defect. Mutations involved PTCH1 (20-40% of cases), PTCH2, suppressor of fused (SUFU) and smo (SMO) genes.1,2 Diagnosis of nevoid basal cell carcinoma syndrome consist of 2 major criteria plus 1 minor criterion or 1 major criterion plus 3 minor criteria.3,4 All our patients fulfilled the diagnostic criteria (Table 1).

<table>
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<th>Major Criteria</th>
<th>Minor Criteria</th>
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<td>≥ 5 BCC or 1 before age 30</td>
<td>Macrocephaly</td>
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<tr>
<td>Odontogenic Keratocysts (Histologically proven)</td>
<td>Congenital malformation: cephalic lip or palate, frontal bossing, coarse facial features, hypertelorism</td>
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<td>≥2 palmar or plantar pits</td>
<td>Preaxial or post axial polydactyly</td>
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<tr>
<td>Lamellar calcification of falx cerebri</td>
<td>Radiologic vertebral or rib anomalies: bifid, splayed, or extra ribs; bifid vertebrae</td>
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<td>First degree relative with Nevoid basal cell carcinoma</td>
<td>Ovarian or cardiac fibroma</td>
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<td>Medulloblastoma</td>
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Table 1 shows the major and minor criteria of NBCC diagnosis.

Common therapies include curettage and electrodessication, cryosurgery, laser ablation, surgical excision, Mohs microsurgery, photodynamic therapy. Topical treatment such as 5% imiquimod cream and 5-fluouracil cream can be considered.1,3 Visnogrel, a hedgehog pathway inhibitor, is recently being approved by FDA for the treatment of advanced BCC and also for patients with NBCC.1,4 All patients should be advised for sun protection.1,4

Recognition of the different clinical presentation of NBCC will allow for early diagnosis and administration of appropriate therapy.

References: