Late Onset of Cardiac Tumour in Naevoid Basal Cell Carcinoma (Gorlin) Syndrome

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Sir,

Naevoid basal cell carcinoma syndrome (NBCCS) is an autosomal dominant disease characterized by developmental abnormalities and predisposition to basal cell carcinomas and various tumours (1). Among them, cardiac tumours appear in 3% of patients (2), mostly in newborns, infants and children. We here report a case of cardiac tumour with a late manifestation, at 59 years of age.

CASE REPORT

A 59-year-old white woman was diagnosed with NBCCS based on multiple basal cell carcinomas of the face, trunk, back and limbs since the age of 15 years, ectopic calcification of the falx cerebri, prognathism due to enlarged jaws, surgery for bilateral ovarian "cysts", and recent surgery for an asymptomatic 4-cm frontal meningioma. There was no family history of NBCCS, but a frameshift mutation of patched (PTCH) gene C942delC was identified on exon 6. The patient had had several episodes of syncope within the past months. Cardiac examination proved normal, without signs of congestive heart failure. Electrocardiography showed sinusoidal rhythm. Cardiac magnetic resonance imaging (MRI) revealed a heterogeneous mass in the myocardium, specifically in the lateral, inferior part of the right ventricular free wall. The mass was iso-intense compared with myocardium on T1-weighted images, hypo-intense on T2-weighted images and heterogeneous on post-contrast images. Focal calcifications of the mass were also noted. MRI pattern was compatible with a benign cardiac fibroma (3). Echocardiography confirmed an echogenic mass of 30×15 mm in the right ventricular free wall without obstruction of outflow. Diastolic and systolic ventricular functions were good. Because of the tumour location and the absence of new episodes, the patient was spared surgery. At 6-month follow-up, she reported no new episodes of syncope.

DISCUSSION

NBBCS is characterized by multiple basal cell carcinomas, palmar and plantar pits, odontogenic keratocysts and various developmental abnormalities (1). Tumours may occur, including meningioma, medulloblastoma and ovarian fibromas. Cardiac fibromas are rare manifestations, affecting 3% of patients (2).

Isolated cardiac fibroma is a rare benign tumour that occurs at all ages, but mainly affects children under 10 years of age (4). NBBCS-associated cardiac fibroma was first reported by Littler (5) in 1979 and comprises 3–5% of all cardiac fibromas (6).

NBCCS-associated cardiac fibroma has the same clinical presentation and pathology as the isolated tumour. Cardiac tumours mostly arise in the left ventricle or septum (4). They can be detected at birth and remain asymptomatic for years (7). Irregular cardiac contour, cardiomegaly, intra-cardiac calcifications on chest X-ray or unexplained cardiac failure and dysrhythmia can reveal the tumour at birth or during childhood (4, 6, 8). To date, approximately 20 cases of cardiac fibroma in NBCCS have been reported, with ages ranging from the prenatal period (4, 8) to 25 years, though most were reported in the first year (4, 6). In a recent review, Gorlin (1) mentioned cardiac fibroma manifesting up to age 60 years. Tumour discovery sometimes leads to retrospective NBCCS diagnosis when personal and family histories are analysed. Prognosis depends on tumour size and location: dysrhythmia, conduction defect or congestive heart failure may lead to death (4, 8). Spontaneous resolutions have been observed (4). Echocardiography is useful for localizing the mass and analysing its functional effect on the heart (4). MRI is the modality of choice for diagnosis, evaluation of myocardial infiltration and local extension, and information regarding surgical treatment (3). Cardiac fibroma presents a specific appearance on MRI that helps in distinguishing it from other cardiac tumours (3). Among these, cardiac metastases of basal cell carcinoma were reported in one case of NBCCS (9). MRI also provides clues for this diagnosis (3). Complete removal of symptomatic tumour is necessary. If radical excision cannot be performed, incomplete resection should be considered. Heart transplantation remains the best treatment for unresectable fibromas but is contra-indicated in NBCCS because of risks related to immunosuppressive therapy. The treatment of asymptomatic patients is still controversial, but the risk of sudden death remains present (6).

Gorlin (10) suggested periodic chest X-rays to evaluate for cardiac fibroma. We believe that any cardiac symptom in a patient with NBCCS should prompt an electrocardiogram and echocardiography to rule out cardiac tumour. Physicians should also systematically examine for NBCCS in cases of isolated cardiac fibroma at any age.

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