Nevoid basal cell carcinoma syndrome- Its management.

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Abstract

Nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin-Goltz syndrome is characterized by basal cell carcinoma (BCC), odontogenic keratocysts (OKC), skeletal, dental, ophthalmic, and neurological abnormalities. As involvement due to this diseases effects large number of organs, a systematic approach to a patient with this disease should be carried out.

Keywords: Gorlin-Goltz syndrome, Odontogenic keratocysts, Corpus callosum, Ectopic calcification.

Accepted on January 31, 2018

Introduction

This syndrome is an autosomal dominant disorder and PTCH1 mutation (tumor suppressor gene) is responsible for this condition. Equal prevalence in males and females have been reported. NBCCS was first described by Gorlin and Goltz in the year 1960. They described this condition to be characterized by a triad which includes multiple BCC, OKC, and bifid ribs. Of the triad mentioned above, OKCs are the most likely to appear first and are usually detected during the first decade of life. Other diseases associated with this syndrome are as follows:

Central nervous system- medulloblastomas, agenesis of corpus callosum, congenital hydrocephalus. Oral cavity- high arched palate, dental anomalies. Skeletal system- anomalies of the rib, spine, bones of the hand and feet. Others- genitourinary tract, cardiac and ophthalmologic anomalies [1].

Eye examination may be overlooked. Ocular findings are in the form of periocular basal cell carcinoma, hypertelorism, strabismus, myelinated nerve fibers, and disorders of the retina and retinal pigment epithelium [2]. First of all, the full history and investigation protocol should be defined [3].

Family history- For any associated disorders with this syndrome. Clinical examinations of the oral cavity, skin, central nervous system, eyes, genitourinary system, cardiovascular system, respiratory system and skeletal system, Genetic testing, Radiographs - Chest, skull, spine, hands, ultrasound of the abdomen for ovarian fibroma. Echocardiogram for cardiac fibroma in children.

Laboratory findings may show an increased serum uric acid level, increased levels of alkaline phosphate and cyclic adenosine monophosphate.

Evans et al. [4] first established major and minor criteria for the diagnosis of this rare entity, later modified by Kimonis et al. [5]. Diagnosis can be established when two major or one major and two minor criteria are present.

Major Criteria

1. More than two BCC or one BCC at younger than 30 years of age or more than 10 basal cell nevi.
2. Any OKC (proven on histology) or polyostotic bone cyst.
3. Palmar or plantar pits (three or more).
4. Falx cerebri calcifications.
5. Bifid, fused, or markedly splayed ribs.
6. Ectopic calcification: Lamellar or early at younger than 20 years of age.
7. Positive family history of nevoid basal cell carcinoma.

Some authors take plurilamellar appearance of the falx cerebri calcification as a pathognomonic symptom of Gorlin-Goltz syndrome.

Minor Criteria

1. Macrocephaly determined after adjustment with height.
2. Skeletal anomalies: hemivertebrae, scoliosis, syndactyly, polydactyly, and shortened 4th metacarpal.
3. Radiological abnormalities like bridging of sella turcica, vertebral anomalies, and modelling defect of hands and feet.
4. Medulloblastoma.
5. Ovarian Fibroma.
6. Congenital malformations: cleft lip or palate, polydactylism or eye anomalies (cataract, coloboma, and microphthalmus).

A Discussion on Diagnosis

The clinical diagnosis relies on specific criteria. Gene mutation analysis confirms the diagnosis. Genetic counseling is mandatory. Antenatal diagnosis is feasible by means of ultrasound scans and analysis of DNA extracted from fetal cells (obtained by amniocentesis or chorionic villus sampling). Some fetuses with NBCCS have large heads and so may need assistance in delivery either by forceps or by Cesarian section. Main differential diagnoses include Bazex syndrome, trichoepithelioma papulosum multiplex and Torre's syndrome

(Muir-Torre's syndrome). Results from several epidemiologic studies have indicated that the risks of BCCs show a strong positive correlation with exposure to ultraviolet (UV) radiation. Thus, these patients need to avoid excess sun exposure. They must use 100% UV protective sunglasses as the skin surrounding the eyes (similar to that of the nose/ears) is vulnerable to BCCs [6].

Conclusion

Treatment modalities for this disease include surgical removal, chemotherapy, laser and photodynamic therapy. The use of vitamin A analogs has also been advocated [7]. Finally, the follow-up schedule for patients suffering from the disease should be specified. Because of aggressive nature and high rate of recurrence, there should be periodic follow-up at regular intervals of 6 months till 5 years, followed by once annually for the entire life [8].

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