VOLUME I/ISSUE I/JUL-DEC 2013 [JOURNAL OF HEAD & NECK PHYSICIANS AND SURGEONS]

LETTER TO EDITOR

GORLIN-GOLTZ SYNDROME- DIAGNOSIS

Hatem Zein Ldeen, R. Malleswar[1]
Department of oral & Maxillofacial Surgery, Cairo , Egypt
1-Chair, Dept of Neurosurgery, NMCH , India

Sir,

Jaw cyst - basal cell nevi or Gorlin-Goltz syndrome [G & G syndrome] is an extremely rare disease inherited by autosomal dominant gene. Etiopathogenesis of this syndrome is due to the abnormalities in the long arm of chromosome 9 (q22.3-q31) and alterations in human PTCH1 gene. The most common findings in the patients with this syndrome include multiple odontogenic keratocysts of jaws and basal cell carcinomas, dental, skeletal, neurologic and ophthalmic abnormalities, ectopic intracranial calcifications of the falx cerebri, and facial dysmorphism. This inherited syndrome may be diagnosed early by an oral & maxillofacial surgeon by routine radiographic investigations in the first decade of life, since the odontogenic keratocysts are usually one of the first manifestations of this syndrome.

In order to arrive at a proper diagnosis of the Gorlin-Goltz syndrome, some diagnostic clinical and radiographic criteria have to be established . Evans et al gave the most important criteria, the presence of major criteria like pigmented baso-cellular carcinomas, Keratocystic Odontogenic Tumors, palmar and/or plantar pits, and ectopic calcifications of the falx cerebri. [1]

Along with these major features of this syndrome , more than 100 minor features have been described in the literature.. The more relevant are the following are ovarian or cardiac fibroma, bifids ribs, macroencephaly, cleft lip and palate. kyphoscoliosis, medulloblastoma, alterations in the sella turcica, lateral displacement of the inner canthus, mandibular prognathia, frontal and biparietal bossing, imperfect segmentation of the cervical vertebrae, rhabdomyosarcoma linfomesenteric cysts that tend to calcify, meningioma ,fibrosarcoma, ocular hypetelorism, short fourth metacarpal, congenital blindness, high arched eyebrows and palate, narrow sloping shoulders, hypogonadism in men, immobile thumbs, low pitch voice in women and renal anomalies. Sometimes, a tall height and even similar features to acromegaly have been associated with the syndrome.[2]

Woolgar et al in 1987 advocated that mean age group for syndromic cases is from 1st to 3rd decade and females are more affected than males. [1]

Treatment choice for this type of syndromic patients with G&G syndrome varies from enucleation to resection and reconstruction considering the site of the lesion, age of the patient , behavior of the lesion , aggressiveness and recurrence of the lesion. [3,4,5]Thorough systematic clinical and radiological examinations along with specific radiographs will help to arrive at a proper diagnosis of the rare syndrome. These investigations prompt an early verification of the disease, which is very important to avoid recurrence of these tumors and ensure better survival rates.

REFERENCES:

1. Evans D, Ladusans E, Rimmer S, Burnell L, Thakker N, Farndon P. Complications of the naevoid basal cell carcinoma syndrome: results of a population based study. Journal of medical genetics 1993;30:460-464
2. Kalogeropoulou C, Zampakis P, Kazantzi S, Kraniotis P, Mastronikolis NS. Gorlin-Goltz syndrome: incidental finding on routine ct scan following car accident. Cases J 2009;2:9087
3. Gailani MR, Bale SJ, Leffell DJ, et al. Developmental defects in Gorlin syndrome related to a putative tumor suppressor gene on chromosome 9. Cell 1992;69:111-117.
4. Jawa DS, Sircar K, Somani R, Grover N, Jaidka S, Singh S. Gorlin-Goltz syndrome. Journal of oral and maxillofacial pathology: JOMFP 2009;13:89.
5. Joshi PS, Deshmukh V, Golgire S. Gorlin-Goltz syndrome. Dental research journal 2012;9:100.

Conflict of Interest – Nil

Acknowledgements – Nil

Support – Nil

Correspondance:

Hatem Ldenn,

Jeddah, Saudi Arabia,

E mail- hatem@gmail.com