FP 9: Basal Cell Carcinoma, Odontogenic Cysts, Brain and Skeletal Abnormalities (Gorlin Goltz) Syndrome in a 46-year-old female

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Abstract:

A 46-year-old woman consulted for a non-healing, necrotic left orbital ulcer that started as a skin-colored, papilla-like lesion on the upper eyelid. There were also hyperpigmented lesions with ill-defined borders over both paranasal areas. Tissue biopsies revealed Basal Cell Carcinoma. Radiologic imaging showed cystic lesions in the mandible, straightening of cervical vertebrae and calcifications of the falx cerebri, tentorium cerebelli, pineal gland and choroid plexus. Based on established major and minor clinical and radiologic criteria, we arrived at a diagnosis of Gorlin Goltz Syndrome or Nevoid Basal Cell Carcinoma Syndrome (NBCCS). She underwent wide excision of the left orbital mass with exenteration, excision of paranasal masses, left total parotidectomy with facial nerve preservation, enucleation of mandibular cyst and cervicofacial reconstruction with skin grafts of the left orbital area and ala.

NBCCS is a rare autosomal dominant disorder with a high tendency for neoplasms and developmental anomalies. Diagnosis can easily be missed if the physician is unaware of its classic but bizarre presentation. Early recognition and prompt specialist referral are very important in order to prevent complications and provide better prognosis. Patients should recognize the importance of follow-up as other presentations of the syndrome may manifest later in life. Family genetic screening and counseling should be undertaken.