SKIN CANCER (OTHER THAN MELANOMA)

NEVOID BASAL CELL CARCINOMA SYNDROME: LITERATURE REVIEW AND CASE REPORT

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Background: Nevoid Basal cell carcinoma syndrome (NBCCS) or Gorlin syndrome is a rare autosomal dominant disease caused by mutations in the human homologue of the Drosophila patched gene PTCH1. Major clinical characteristics include an early development of multiple basal cell carcinomas (BCCs), odontogenic keratocysts of the jaw, palmar and plantar pits, lamellar calcification of the falx cerebri, and family history. Minor manifestations include, among others, craniofacial, vertebral and other skeletal anomalies, medulloblastomas and ovarian or cardiac fibromas.

Observation: A 65 years old female patient attended our outpatient department for multiple BCCs on the face. She described the appearance of BCCs since the age of 24. Surgical excision of 15 BCCs had already been conducted in the past. An odontogenic keratocyst had been removed surgically at the age of 16. Her brother and children suffer from the same symptoms. According to the clinical presentation and taking into account her medical history, the diagnosis was nevoid basal cell carcinoma syndrome and the treatment of our choice was vismodegib. 6 months later, there is only one BCC left in the face, whose size has been remarkably reduced.

Key message: NBCCS is a rare disease but always to be suspected in the presence of an early onset of multiple BCCs. Correct diagnosis should be set as soon as possible in order to choose the effective treatment. Vismodegib, a small molecule inhibitor of smoothened (SMO) which blocks the activation of the sonic hedgehog pathway, has been successfully administrated lately in treatment of BCCs. However, further carefully designed studies are required to confirm its efficacy and to propose alternative treatments for NBCCS.