SKIN CANCER (OTHER THAN MELANOMA)

VISMODEGIB HEDGEHOG-SIGNALING INHIBITION AND TREATMENT OF BASAL CELL CARCINOMAS IN GORLIN-GOLTZ SYNDROME

S. R. Mendes [1] - A. Brinca [1] - R. Vieira [1]

Centro Hospitalar E Universitário De Coimbra, Dermatology Department, Coimbra, Portugal [1]

Background: The Gorlin-Goltz syndrome is an infrequent multisystemic disease inherited under a dominant autosomal pattern, with several diagnostic criteria, but only two major and one minor criteria or one major and three minor criteria are required to the diagnosis. It is characterized by keratocystic odontogenic tumors in the jaw, multiple basal cell carcinomas, skeletal abnormalities and central nervous system anomalies (medulloblastoma).
Vismodegib is an oral inhibitor of the hedgehog-signaling pathway with potential for reducing the burden of multiple basal cell carcinomas and jaw keratocystic odontogenic tumors.

Observation: We present the case of a 25-year-old patient with Gorlin-Goltz syndrome with past history of medulloblastoma at the age of 2 years treated with surgery and radiotherapy. Initially, he was observed in maxillofacial service due to multiple odontogenic keratocyst. The first basal cell carcinoma was excised in the cervical region at the age of 16 years. From then he was observed in our department with high count of basal cell carcinomas that made the surgical treatment impossible for curative purposes. Vismodegib was started, and a clinically complete response was achieved at week 16, confirmed with skin biopsy in the site of the larger lesion. Adverse effects included diffuse alopecia and muscle spasms, reverted with the interruption of treatment at week 20. Recurrence was observed after 4 months. Recurrent disease responded to the reintroduction of the drug. The patient is well controlled with a on-off regimen of vismodegib ongoing for more than 3 years (reintroduction after recurrence and discontinuation after a complete response is achieved).

Key message: Vismodegib seems to be effective in basal cell carcinomas associated to Gorlin-Goltz syndrome when surgical excision is not the best choice. The efficacy seems not to be affected by an on-off therapeutical schedule.