SKIN CANCER (OTHER THAN MELANOMA)

HAPPLE TINSCHERT SYNDROME

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BACKGROUND: Happle-Tinschert Syndrome is a multisystem disease characterized by segmentally arranged cutaneous basaloid follicular hamartomas associated with ipsilateral osseous, dental and cerebral abnormalities including tumours. Mutations in SMO have been described.

CASE HISTORY: A 54-year-old man was born with with right side fascial asymmetry and mild macrocephaly.
At 26 he developed his first basal cell carcinoma. Subsequently he has developed numerous basal cell carcinomas over the right upper quadrant of his body. Examination showed the right side of his face was smaller than the left. A vertical line ran down the centre of the nose and on to the philtrum. He had large pits on his right palm and focal linear hypoplasia on the third and fourth fingers of the right hand. The right thumb had a coarse linear streak. There were numerous shiny papules lying over the right upper quadrant of his body. Histology of the papules showed basal cell carcinoma. X-rays of his jaw and ribs were normal.
A tentative diagnosis of segmental basal cell naevus syndrome was made.
He was given sun care advice, vitamin D and treated with excision, curettage and cautery, cryotherapy, photodynamic therapy and imiquimod. Genetic testing of blood did not show a PTCH mutation. Testing of tissue from two basal cell carcinomas showed a weak PTCH deletion in one sample. This was insufficient to confirm a diagnosis of segmental basal cell naevus syndrome.
The diagnosis was reviewed and the histology re-examined, looking for basaloid follicular harmatoma, which would suggest Happle-Tinschert syndrome.
The histology showed multiple basaloid follicular hamartomas, with basal cell carcinoma arising from them.

KEY MESSAGE: Happle-Tinschert syndrome (HTS) should be considered in the differential of segmental basal cell naevus syndrome. The diagnosis can be confirmed by identifying basaloid follicular harmatoma on histology.