Multiple jaw cysts and ectopic calcification

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DESCRIPTION

A 41-year-old woman presented with pus discharge from the gingival pocket and a 10-year history of intermittent buccal swelling on the left side. Physical examination revealed increased height, macrocephaly, hypertelorism, wide nasal bridge and sloping shoulders. Orthopantomography showed multiple jaw cysts (figure 1). Keratocystic odontogenic tumour (KCOT) was histologically suggested by the preoperative biopsy. CT revealed ectopic calcifications in the falx and tentorium (figure 2A). Subcutaneous radiopaque bodies that seemed like granules were diffusely scattered in the skull (figure 2B). Punch biopsy revealed foreign

calcifications but no histological abnormalities in the hair follicle. Then all jaw cysts were extirpated under general anaesthesia. The histological analysis confirmed the diagnosis of KCOT. These clinical presentations were consistent with a diagnosis of nevoid basal cell carcinoma syndrome (NBCCS). On the other hand, palmar or plantar pits, basal cell carcinoma (BCC) and bifid ribs, which are important diagnostic criteria,¹ were not noted. The diagnosis of NBCCS was finally established by the genetic analysis of *PTCH1*. Thus BCC development and cyst recurrence should be carefully observed.

Learning points

- ▶ Nevoid basal cell carcinoma syndrome (NBCCS) is a rare syndrome that is characterised by keratocystic odontogenic tumours in the jaw, lamellar calcification of the falx, palmar/plantar pits and basal cell carcinomas.¹
- ▶ Genetic analysis of *PTCH1* is an alternative method to establish the diagnosis of NBCCS.
- ▶ Subcutaneous calcification in the skull is scarcely observed even in NBCCS.

Figure 1 Panoramic radiography of the jaw demonstrating multiple cysts.

Figure 2 Ectopic calcifications in the falx and tentorium (A) and subcutaneous radiopaque bodies in the skull (B).

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Patient consent Obtained.

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