# A subcutaneous neck lump in a young man

Ultrasound-guided fine needle aspiration

Tony Maddox, consultant cellular pathologist

West Herts Teaching Hospitals NHS Trust

#### Clinical presentation

- 36 yo male
- 1x2cm lump left side of neck, present for several months
- Just behind left ear and hairline
- Father had history of lymphoma
- Seen by ENT and referred for ultrasound imaging



#### Ultrasound appearances

- Ovoid, 1.4cm
- Subdermal/subcutaneous
- Sharply defined, heterogeneously hypoechoic
- Some hyperaemia (increased vascularity) in surrounding tissue
- Diagnosis ?epidermal cyst
- FNA performed

NB. "Hypoechoic" means little ultrasound echo back from the target, ie looks black. "Hyperechoic" means lots of echo, ie looks white and speckly. Epidermis (the very top layer of skin) and adipose (fatty) tissue are hyperechoic normally. The dermis (between epidermis and subcutaneous adipose tissue) is hypoechoic normally.





# Low power (x10) view



"Busy". Large aggregates of cells. Orangophilia, possibly keratinisation, in Pap



#### X20 view - Pap



Large aggregates of cells with further confirmation of probable keratinisation (arrows)



#### X40 view

Some of the aggregates are composed of basaloid-appearing cells with prominently-nucleolated nuclei. There are also multinucleated cells (arrow) and a orange structure in the Pap with an apparent hole in the middle (arrowhead)







BAC British Association for Cytopathology

#### X40 view



Some of the multinucleated cells are large (top) and are associated with the basaloid cells (middle)



#### And at higher power...



Some more of the rather "crinkly" structures that are orange in the Pap and how they look in the MGG. In fact, they are easier to find in the Pap whereas, in the MGG, they blend into the background



# What's your diagnosis?



# Pilomatrixoma

- Also known as:
  - Pilomatricoma
  - Calcifying epithelioma of Malherbe



#### Pilomatrixoma - cytopathology

- Variable proportions of
  - Basaloid cells may have distinct nucleoli
  - Shadow/ghost cells (the crinkly things) pale blue/grey in Romanowsky stains, pale orange/brown in Pap. Often easier to distinguish in Pap
  - Anucleated squamous cells
  - Multinucleated giant cells
  - Calcification



# Pilomatrixoma

- Benign skin adnexal tumour derived from hair matrix
- Most occur in children (median age 16) but have been reported at any age
- Marginally more common in women than men
- Usually solitary. Occasionally multiple and then may be associated with one of a number of rare syndromes
- Sites:
  - Most commonly head and neck (~50%)
  - Then upper and lower extremities
  - Then trunk
- Most associated with mutations in CTNNB1 gene (gene product is beta catenin, part of Wnt signalling pathway, important in hair shaft differentiation)



# Pilomatrixoma – typical presentation

- Slow-growing, firm, sometimes hard mobile dermal nodule
- Sometimes so hard that they show a "see-saw" (or in the US, "teeter-totter") sign where pressing down on one end of the lesion causes the other to rise
- 0.5 5cm
- Flesh-coloured or white but may have overlying blue or pink tinge
- Usually asymptomatic but may discharge or be tender or itchy, very occasionally ulcerate
- Often misdiagnosed as epidermal cyst



**J Chin Med Assoc** 2004;67:633-636

Chih-Chieh Chuang Hou-Chun Lin Case Report

Pilomatrixoma of the Head and Neck



# Pilomatrixoma - ultrasound

- Well circumscribed
- Junction of deep dermis and subcutis
- Characteristically hyperechoic centre and hypoechoic periphery
  - "doughnut" pattern
- May be entirely hypoechoic



# Pilomatrixoma - histopathology

- Lobulated, well-circumscribed
- Sheets of basaloid cells abrupt keratinisation
- Ghost/shadow cells
  - Not just featureless, necrotic, cell-sized "blobs" but have granular, pale pink cytoplasm (in H&E) and either a hole or a "faded" nucleus where nucleus used to be
- May be numerous multinucleated cells (reaction to keratin)
- May be calcification and, rarely, true ossification
- Basaloid cells (nuclei and cytoplasm) immunopositive for beta catenin



# Pilomatrixoma – classical histopathology



# Pilomatrixoma - histopathology



Basaloid cells

#### Pilomatrixoma - histopathology

Basaloid cells



"Ghost cells" (right) and multinucleate giant cells





#### Pilomatrixoma – behaviour and treatment

- Excision is almost always curative
- 1-2% of cases recur after incomplete excision
- Some lesions show acceleration in growth ("proliferative") while still being histopathologically benign
- Very rarely, genuine malignant transformation (pilomatrix carcinoma) occurs, usually in more elderly patients and probably in pre-existing benign lesions



# Cytopathology of pilomatrixoma – key points

- Clinical details important:
  - (usually) young age
  - (usually) head and neck
  - location in deep dermis/subcutis boundary
- Most sensitive findings for diagnosis on FNA (in order)
  - Ghost/shadow cells
  - Basaloid cells
  - Giant cell reaction



#### References

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