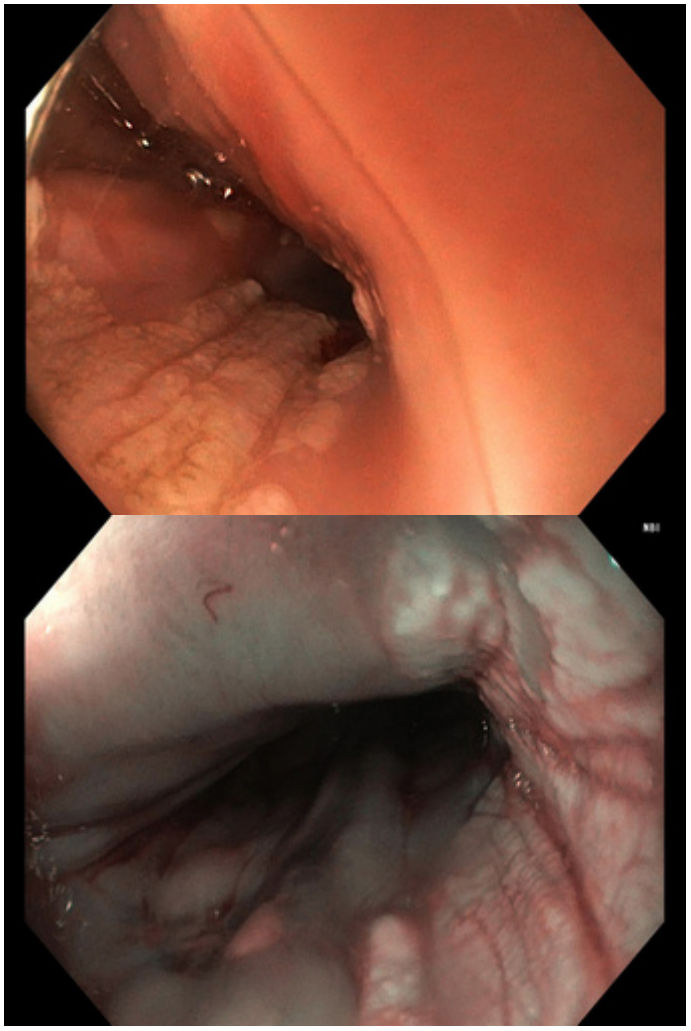
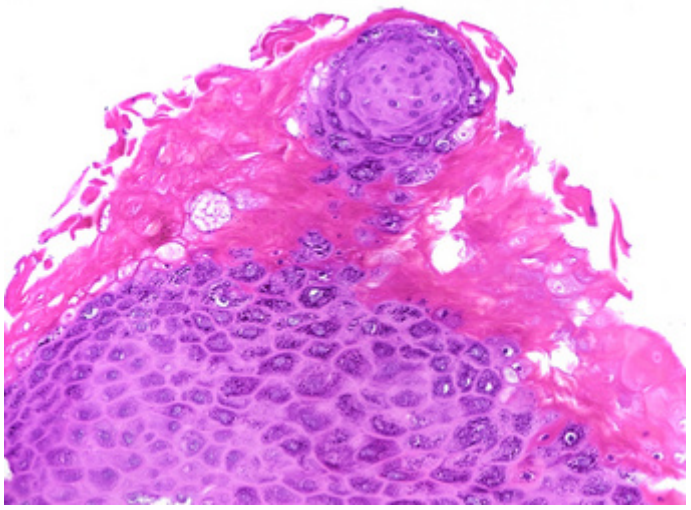
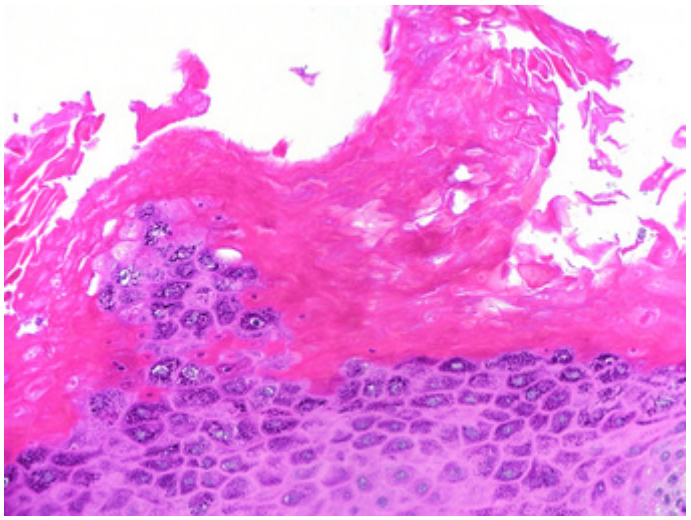
March 2019

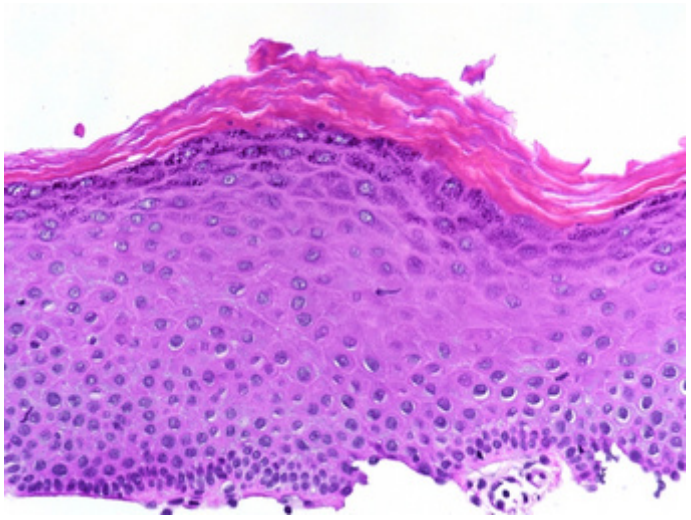
Oesophageal biopsy of an 80-year-old male with symptoms of gastro-oesophageal reflux disease.

What is your diagnosis?









Diagnosis:

Epidermoid metaplasia.

Comment:

Native and narrow-band endoscopic imaging revealed reflux changes at the gastro-oesophageal junction (Los Angeles A), but also sharply demarcated areas of whitish, slightly elevated patches covering the mucosa of the mid and distal oesophagus [Panels A and B]. Biopsies taken from the lesion demonstrated stratified squamous epithelium with mild acanthosis, yet marked hypergranulosis and compact hyperorthokeratosis [Panels C and D]. There were no signs of (hyper-)regeneration, that is, the basal cell layers appeared normal [Panel E] and there was no papillary elongation. Dysplasia was likewise absent.

Oesophageal epidermoid metaplasia is a rare finding, which corresponds to the endoscopic diagnosis of “oesophageal leukoplakia”. In a series of 1048 consecutive oesophageal biopsies the incidence was found to be only 0.19%. The disease, which on the histological level largely resembles normal epidermis, is mainly diagnosed within the mid- to distal portions of the oesophagus. Elderly individuals presenting with dysphagia or reflux symptoms are mainly affected. Severe cases may present as diffuse hyperkeratosis of the entire organ (“crackleware” oesophagus).

Differential diagnosis includes pill oesophagitis, corrosive damage, sloughing oesophagitis, rare inherited keratinization defects (such as tylosis) and mucosal hyperkeratosis syndromes, as well as lichen planus.

The aetiology is not fully established, although association with exogenic toxins, such as smoking and heavy alcohol consumption has been described. Epidermoid metaplasia is regarded as a precursor lesion to squamous dysplasia and carcinoma, the risk of progression is however largely unclear. Clinical and

endoscopic follow-up is recommended, but surveillance intervals still need to be defined (owing to the rarity of disease no formal guidelines exist). It may be of note that the patient described by our group in 2009 (Kieninger et al.) developed cancer eight years later, resulting in oesophagectomy. She is however fine since then.

For further reading:

- Westerterp M, Busch OR, Bergman JJ, et al. A "crackleware" oesophagus. J Clin Pathol. 2005; 58: 1325-1327.
- Kieninger EM, Siebert F, Langner C. Endoscopic treatment of spontaneous, incomplete esophageal rupture in a patient with "crackleware esophagus". Endoscopy. 2009;41 Suppl 2:E123-E124.
- Singhi AD, Arnold CA, Crowder CD, et al. Esophageal leukoplakia or epidermoid metaplasia: a clinicopathological study of 18 patients. Mod Pathol. 2014; 27: 38-43.
- Cottreau J, Gruchy S, Kamionek M, et al. Prevalence of oesophageal epidermoid metaplasia in 1048 consecutive patients and 58 patients with squamous neoplasms. Histopathology. 2016; 68: 988-995.
- Singhi AD, Arnold CA, Lam-Himlin DM, et al. Targeted next-generation sequencing supports epidermoid metaplasia of the esophagus as a precursor to esophageal squamous neoplasia. Mod Pathol. 2017; 30: 1613-1621.

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