



**Bioptická** laboratoř

CYTOLOGIE | BIOPSIE | MOLEKULÁRNÍ GENETIKA



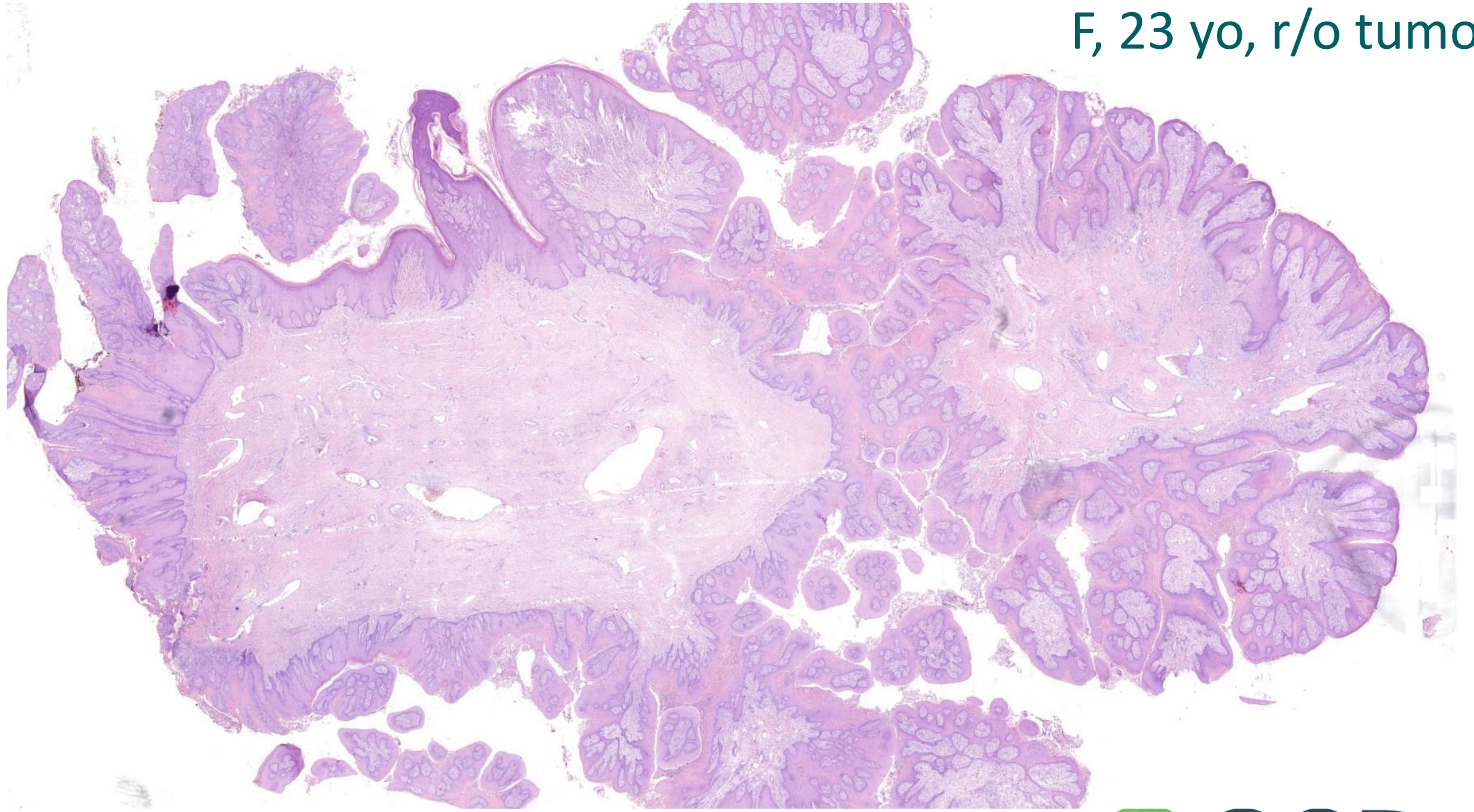
**LETNÝ BIOPTICKÝ SEMINÁR SD-IAP**  
**Rare genodermatosis in routine  
pathologist's practice**

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CSD Medical Laboratory, Kyiv, Ukraine



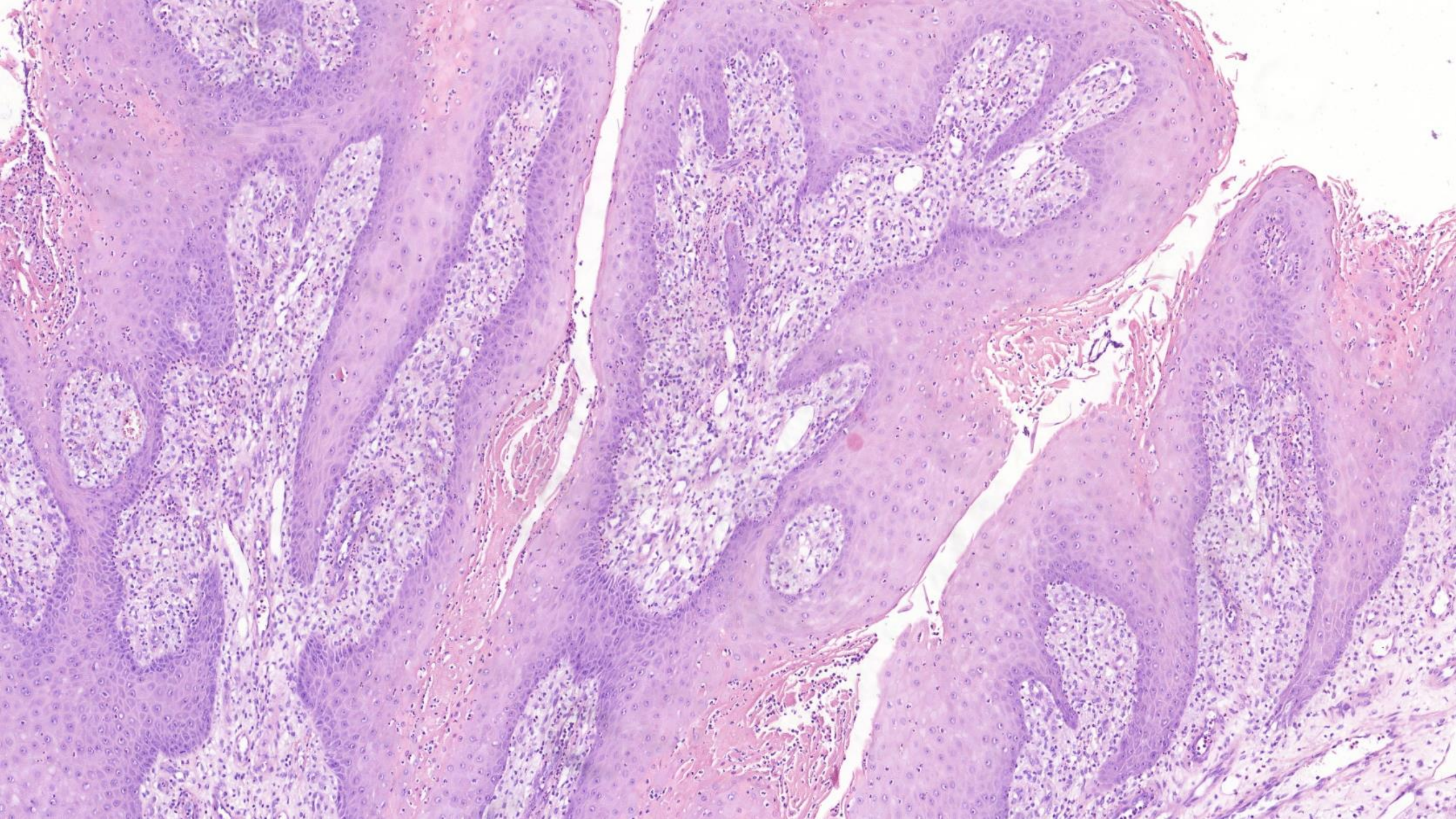


F, 23 yo, r/o tumor

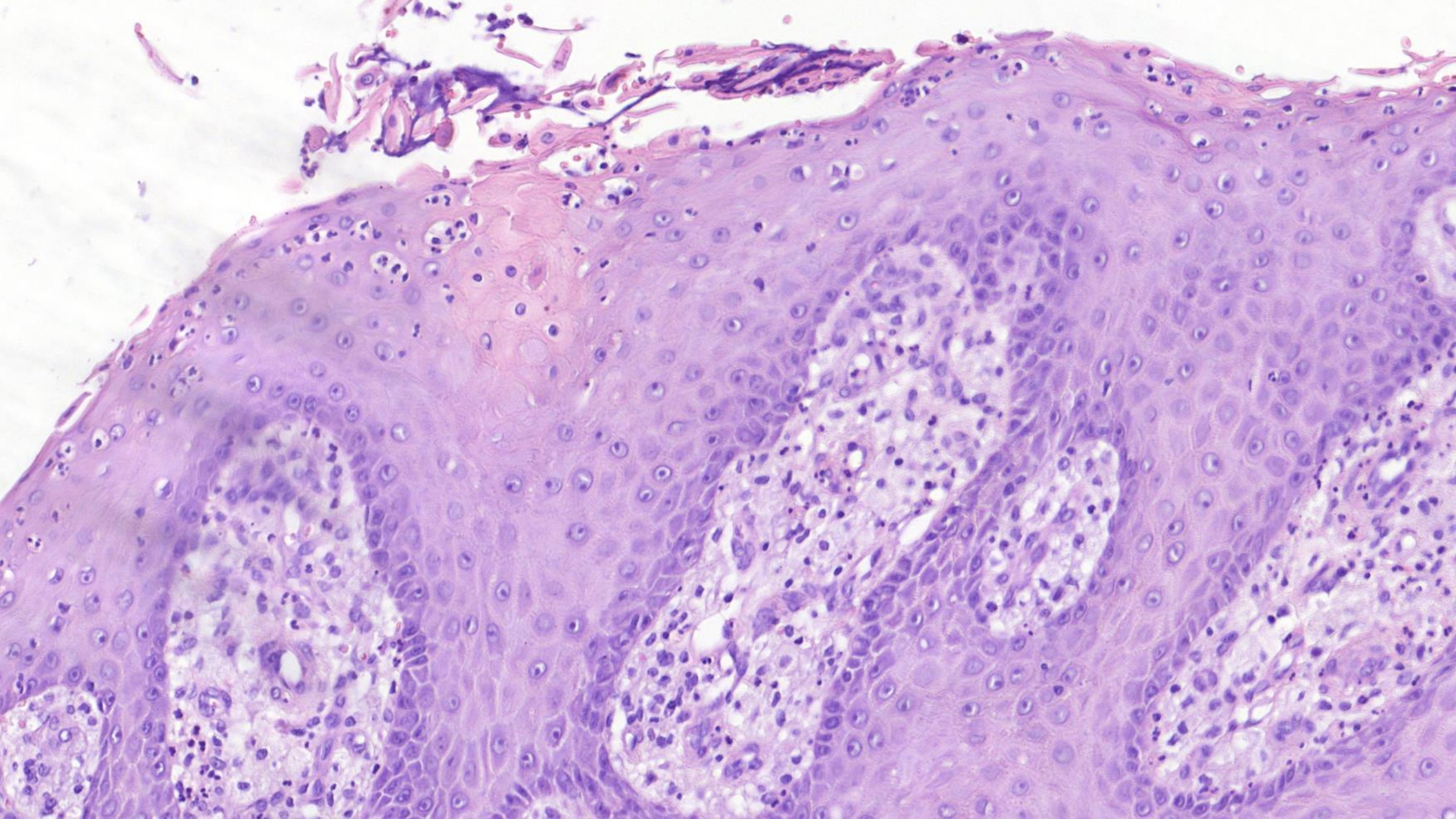


Clinical case 1











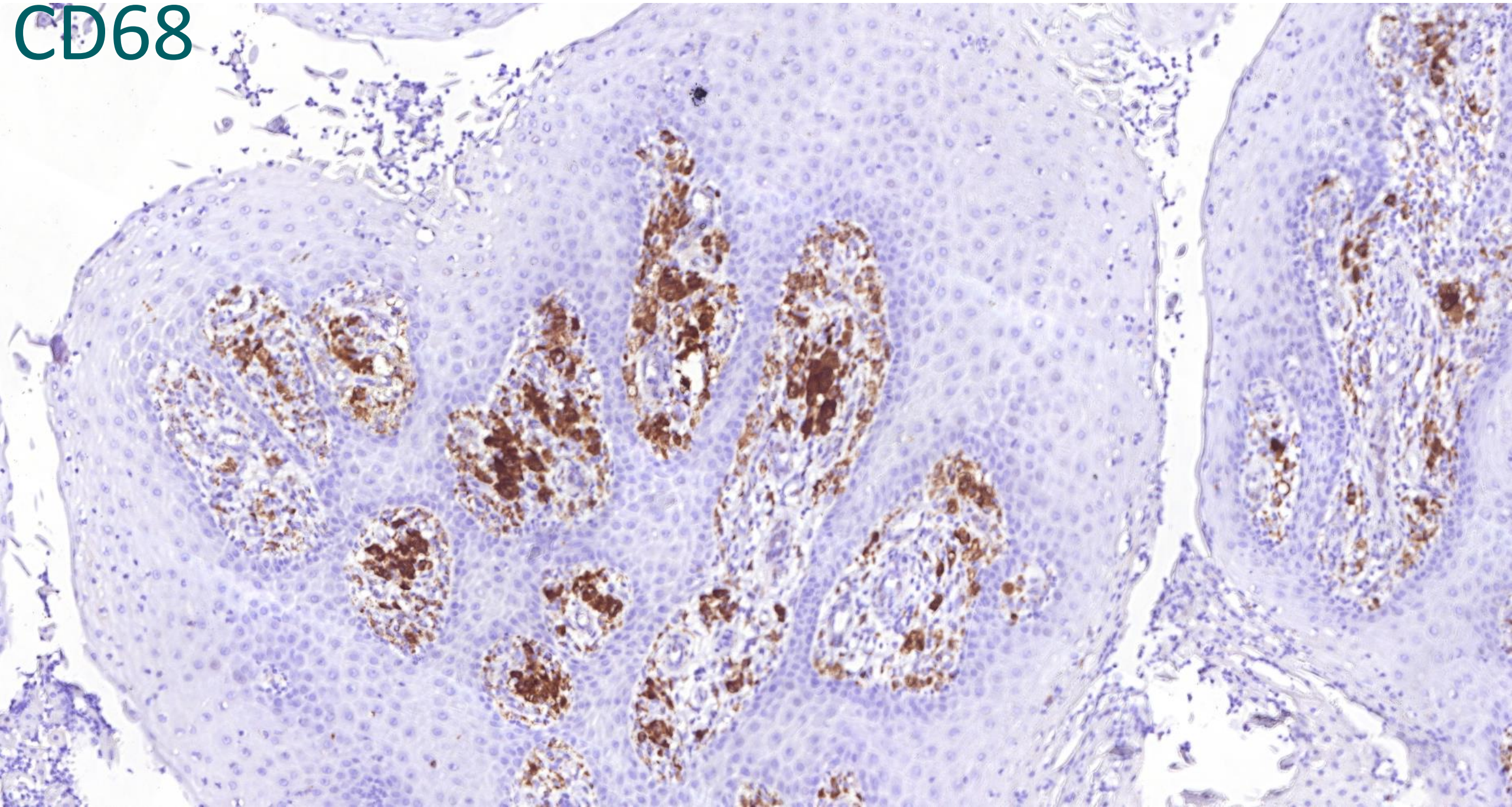


## Your diagnosis?

- Inflammatory linear verrucous epidermal naevus or ILVEN
- CHILD syndrome – associated Verruciform Xanthoma
- Verruciform Genital-Associated Xanthoma
- Irritated fibroepithelial polyp

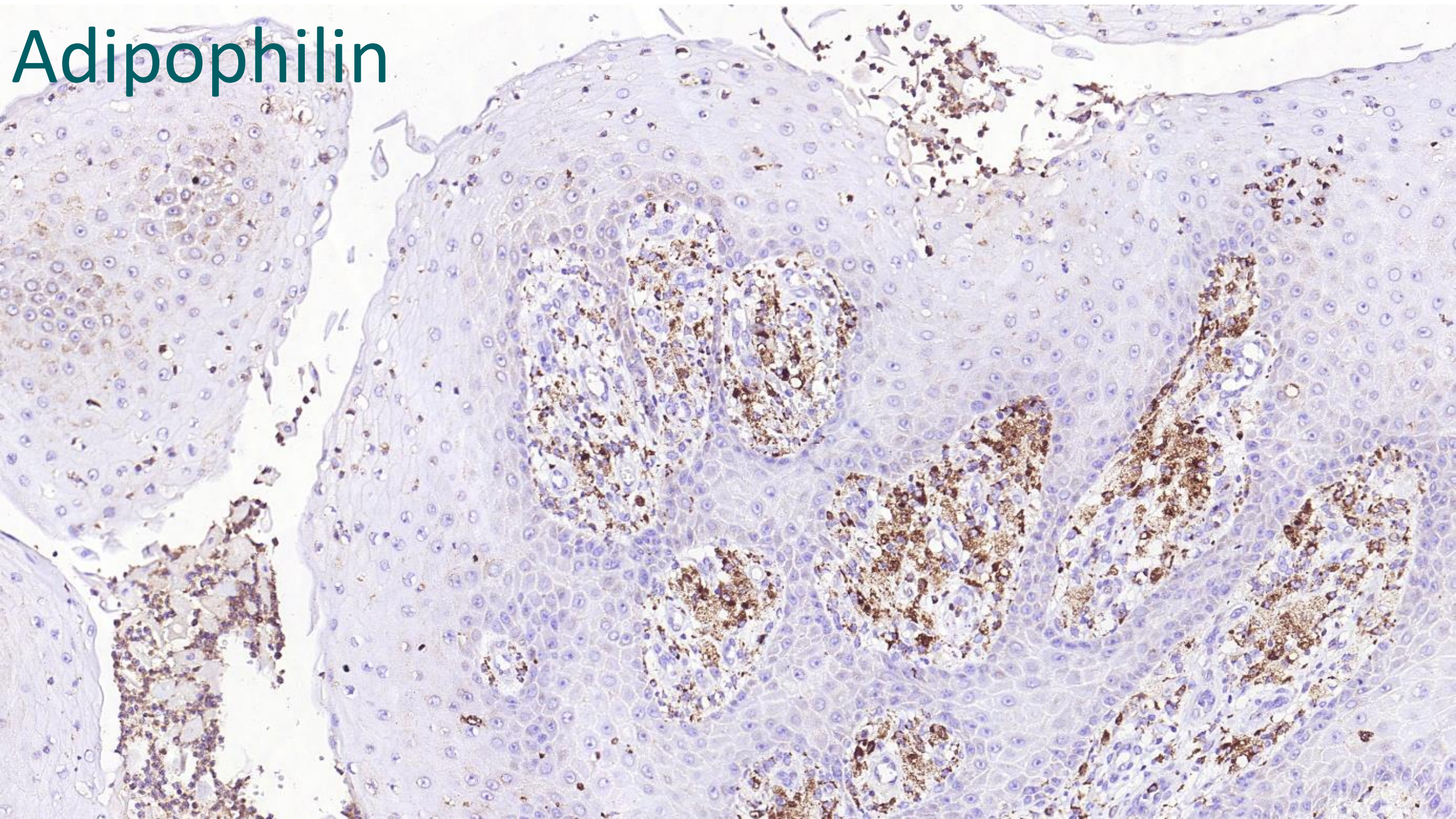


CD68





# Adipophilin







# Clinical case 1



- A 23-year-old girl was admitted to the hospital with linear lesions arranged across Blaschko's lines, running from the pectoralis region to fingertips, also groin, and inner thigh, leg, and dorsum of the foot were affected, prominent nails involvement, slowly growing tumorous lesions in the vulvar area and lower extremities with severe bleeding.
- Clinical diagnosis on the time of admission was r/o SCC arising in IVLEN

























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# Verruciform xanthomas

- Verruciform xanthomas are benign, asymptomatic wart-like lesions most commonly found in the oral cavity.
- Extra-oral sites were also reported such as the penis, scrotum, and vulva.
- The age of onset of the female genital lesions ranges from shortly after birth [in association with congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD syndrome)] to age 85 years.
- Pathologic features include hyperkeratosis with parakeratosis, acanthosis, elongated rete ridges, neutrophilic inflammation in the dermis, and foam cells in the dermal papillae.





# Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects or CHILD syndrome

- Unilateral involvement
- Sharp midline demarcation of congenital ichthyosiform or psoriasis-like lesions
- Skin lesions arranged in a linear fashion following Blaschko's lines
- Ptychotropism – high affinity to body folds
- Nail dystrophy
- Multiple verruciform xanthomas





# Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects or CHILD syndrome (OMIM: 308050)

- CHILD syndrome is an acronym for an X-linked dominant disorder characterized by congenital hemidysplasia with ichthyosiform erythroderma and limb defects. The mutations are lethal in hemizygous males (**Happle et al., 1980**).
- **Falek et al. (1968)** described sibs with this combination
- **Happle et al. (1980)** used the acronymic designation 'CHILD syndrome'
- CHILD syndrome is observed only in females, **Happle et al. (1996)** described the case of a 2-year-old boy of Egyptian origin born of healthy unrelated parents. One more boy was describe recently.





# CHILD syndrome. Pathophysiology

- CHILD syndrome is inherited in an X-linked dominant fashion and involves a mutation in the NSDHL (NAD[P]H steroid dehydrogenase–like protein) gene.
- The gene has been localized to Xq28 and encodes for 3beta-hydroxy sterol dehydrogenase, which catalyzes a step in the cholesterol biosynthetic pathway.
- This enzyme is located both within the membranes of the endoplasmic reticulum and on the surface of intracellular lipid storage droplets.
- Several different types of mutations in the gene have been documented, including missense, nonsense, and stop mutations, all resulting in a loss of function of NSDHL.
- Clinical variations in the extent of involvement are not thought to be secondary to the specific type of mutation, but rather the differences in the pattern of X inactivation.
- The striking laterality of the syndrome may arise from this impaired cholesterol processing, causing abnormal sonic hedgehog signaling, which, in embryogenesis, is important in spatial patterning.





## Clinical case 2



- A 9-year-old girl presented with unilateral left-side waxy-like plaques that were arranged across Blaschko's lines
- Limb abnormality was also present: absence of third toe
- Presence of verrucous tumor of the vulva
- Clinical diagnosis: CHILD syndrome





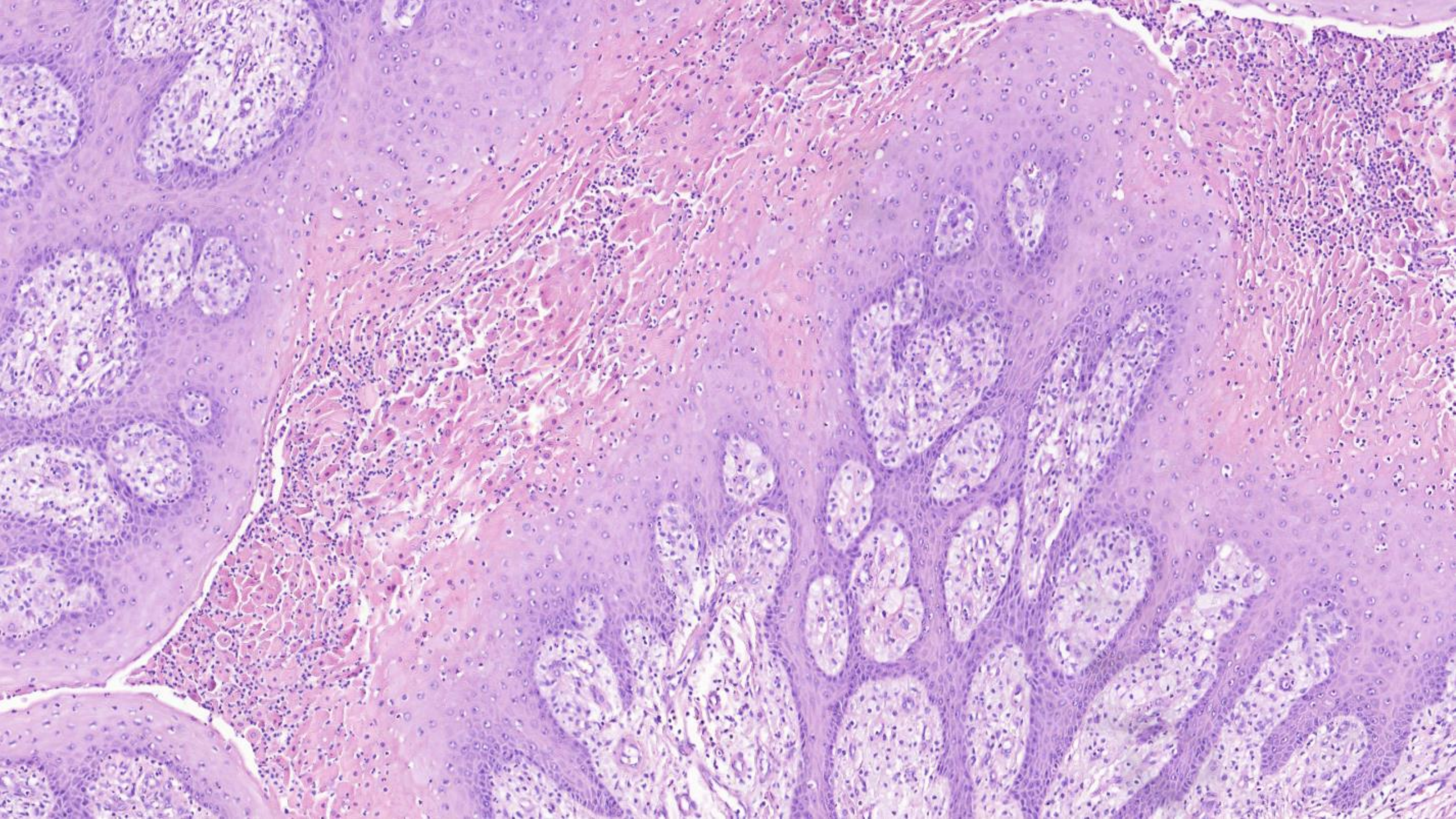
















# Diagnosis

- Diagnosis of CHILD syndrome was proved according to typical clinical features
- Histological findings were corresponded with verruciform xanthoma
- Only 5 cases of left-side involvement reported in the world literature

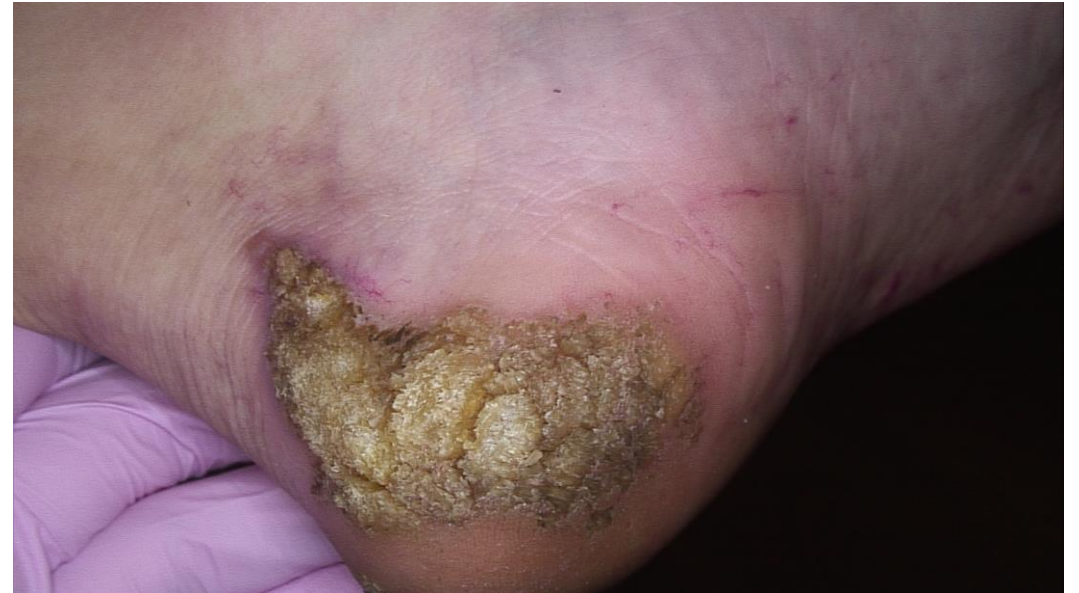




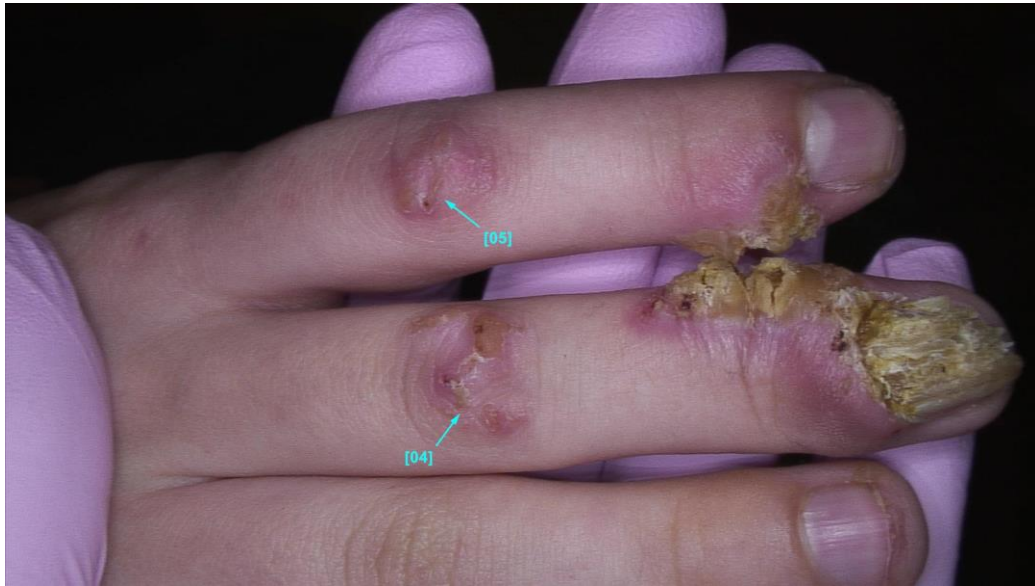
## Clinical case 3

- A 19-year-old female was presented to the dermatological department with multiple skin lesions that affected her hands and feet. Also it was a verruciform tumor in her anogenital region.
- Diffuse nonscarring alopecia was presented
- Punch biopsies of her hands and vulva lesions were performed.
- Clinical diagnosis at the time of admission: epidermodysplasia verruciformis, HPV







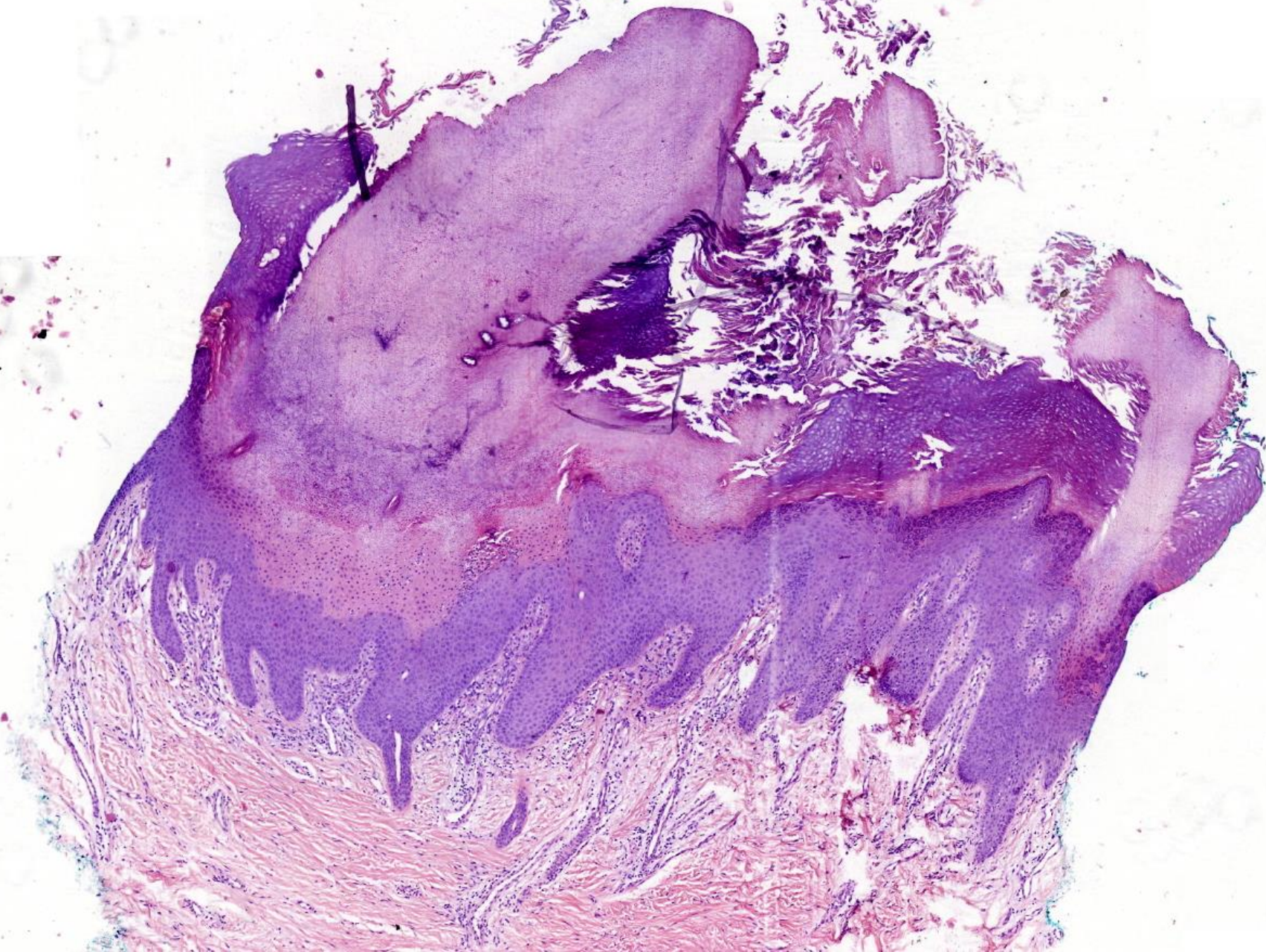






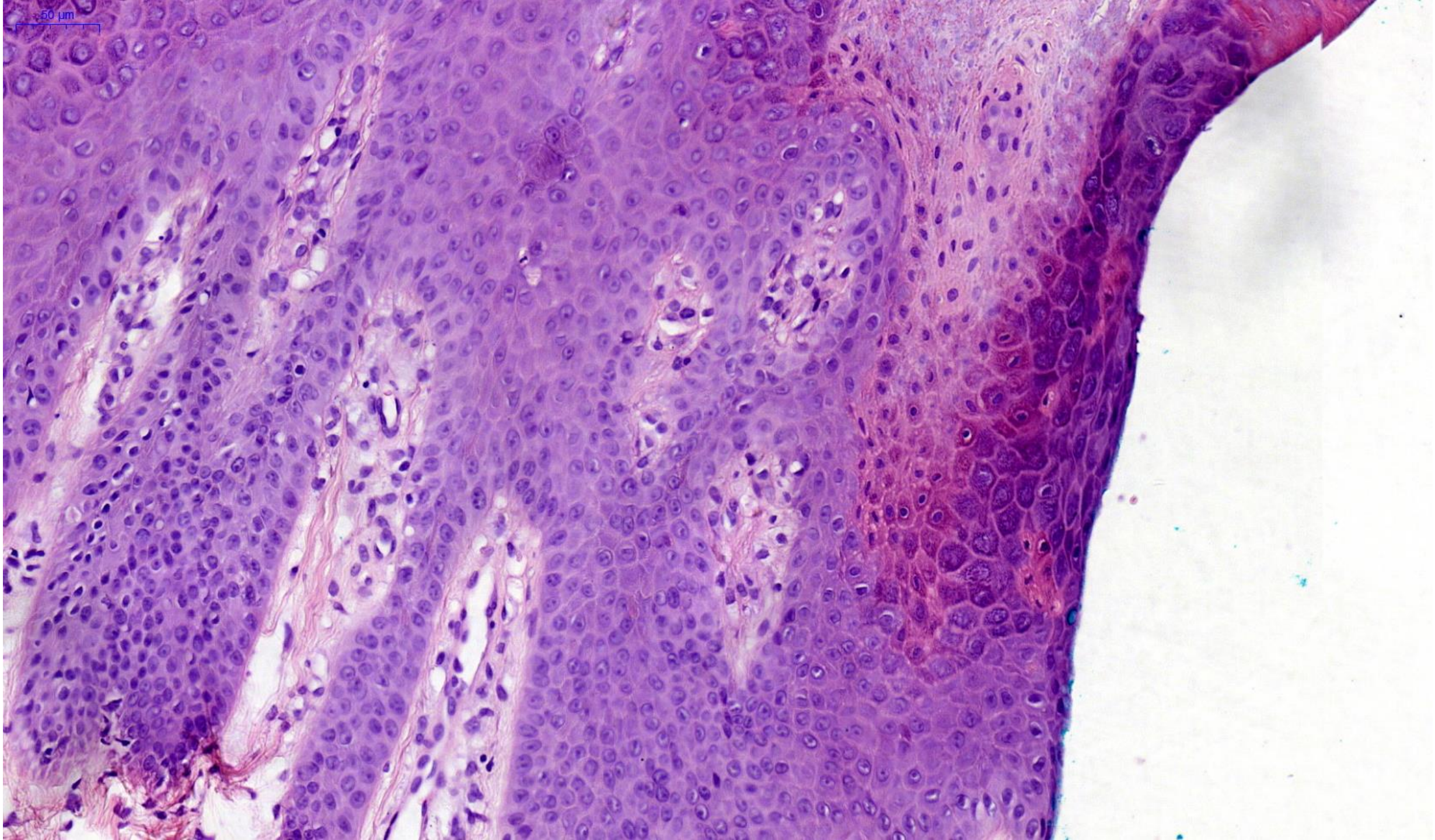


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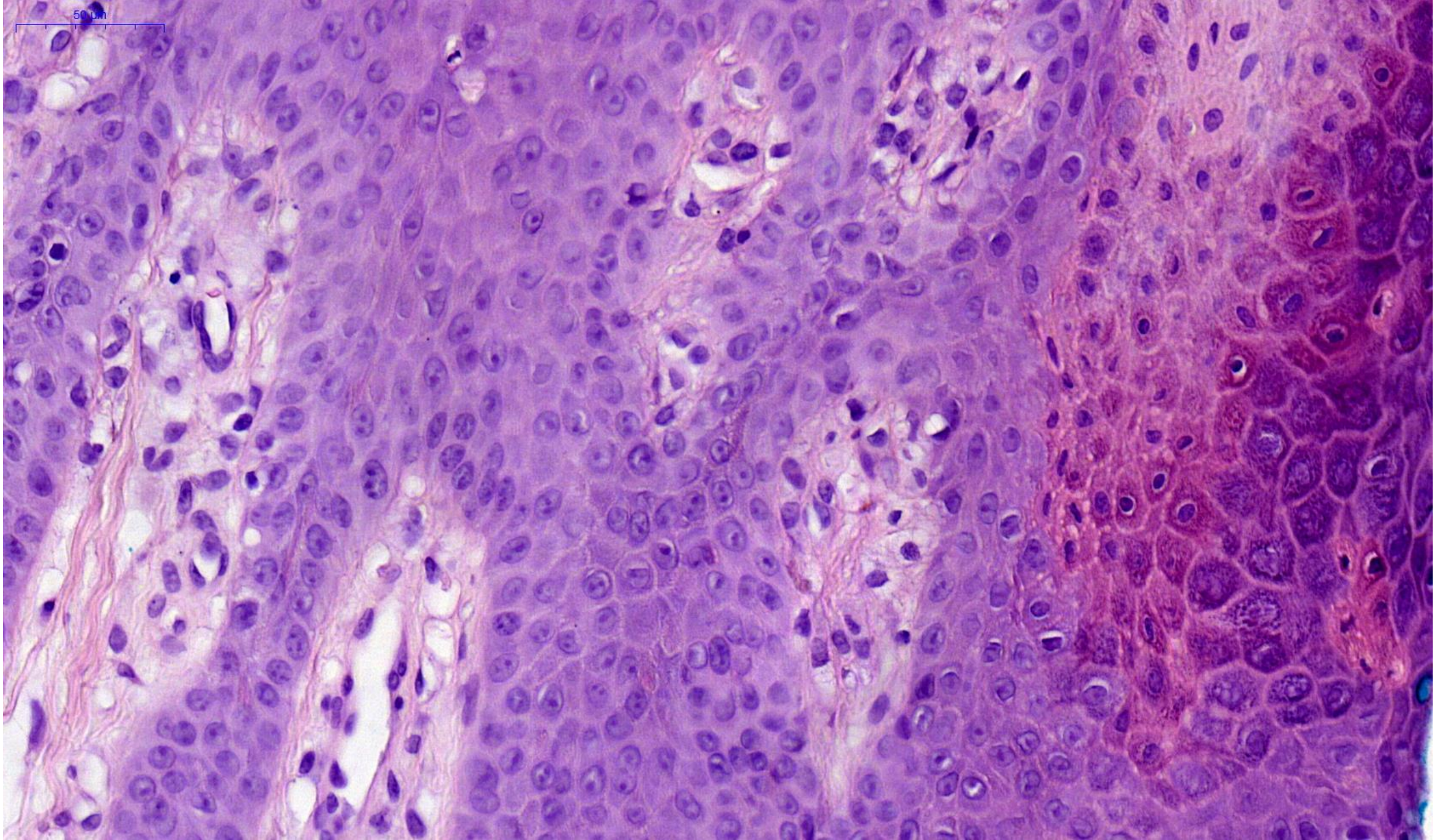


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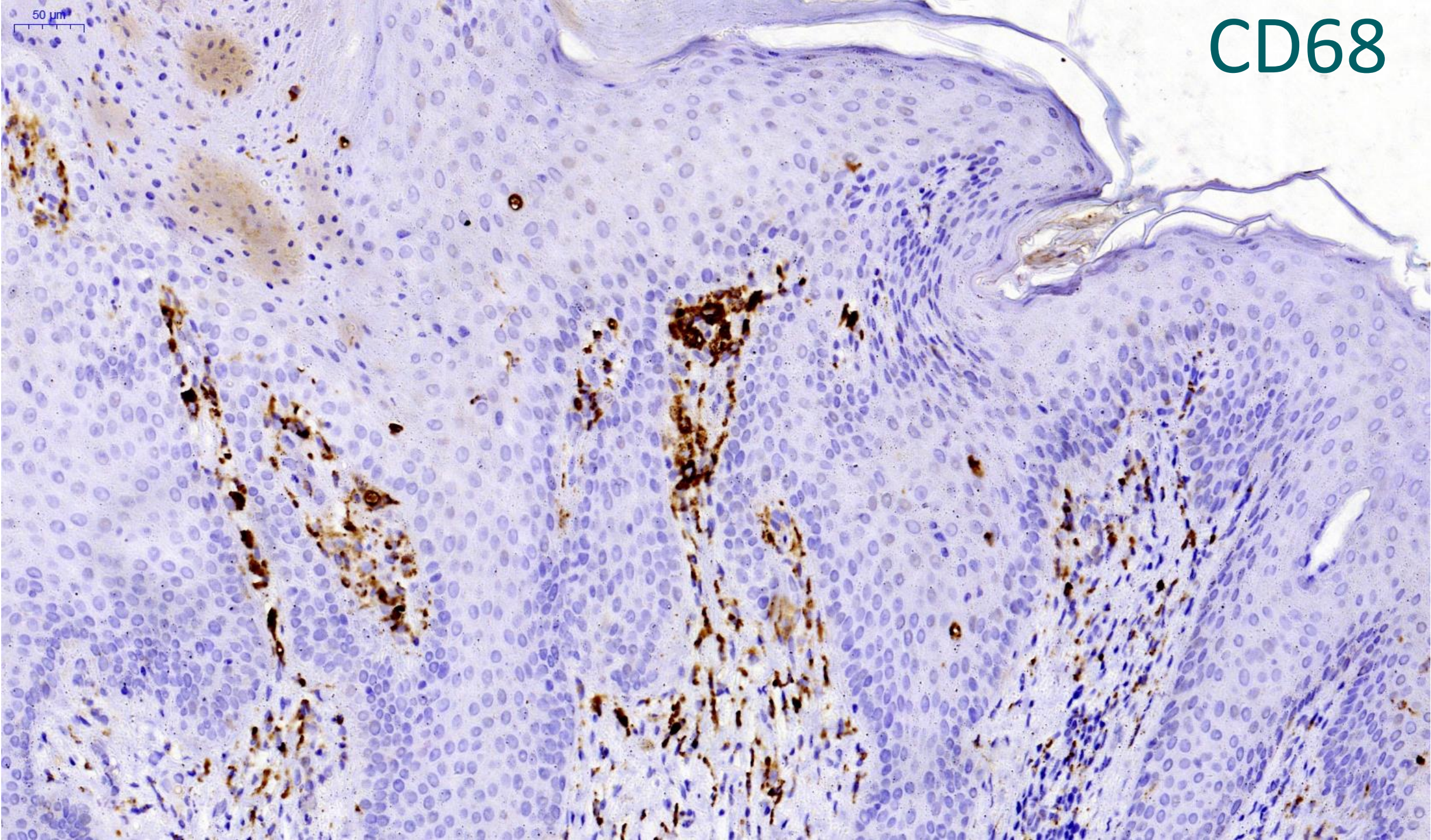
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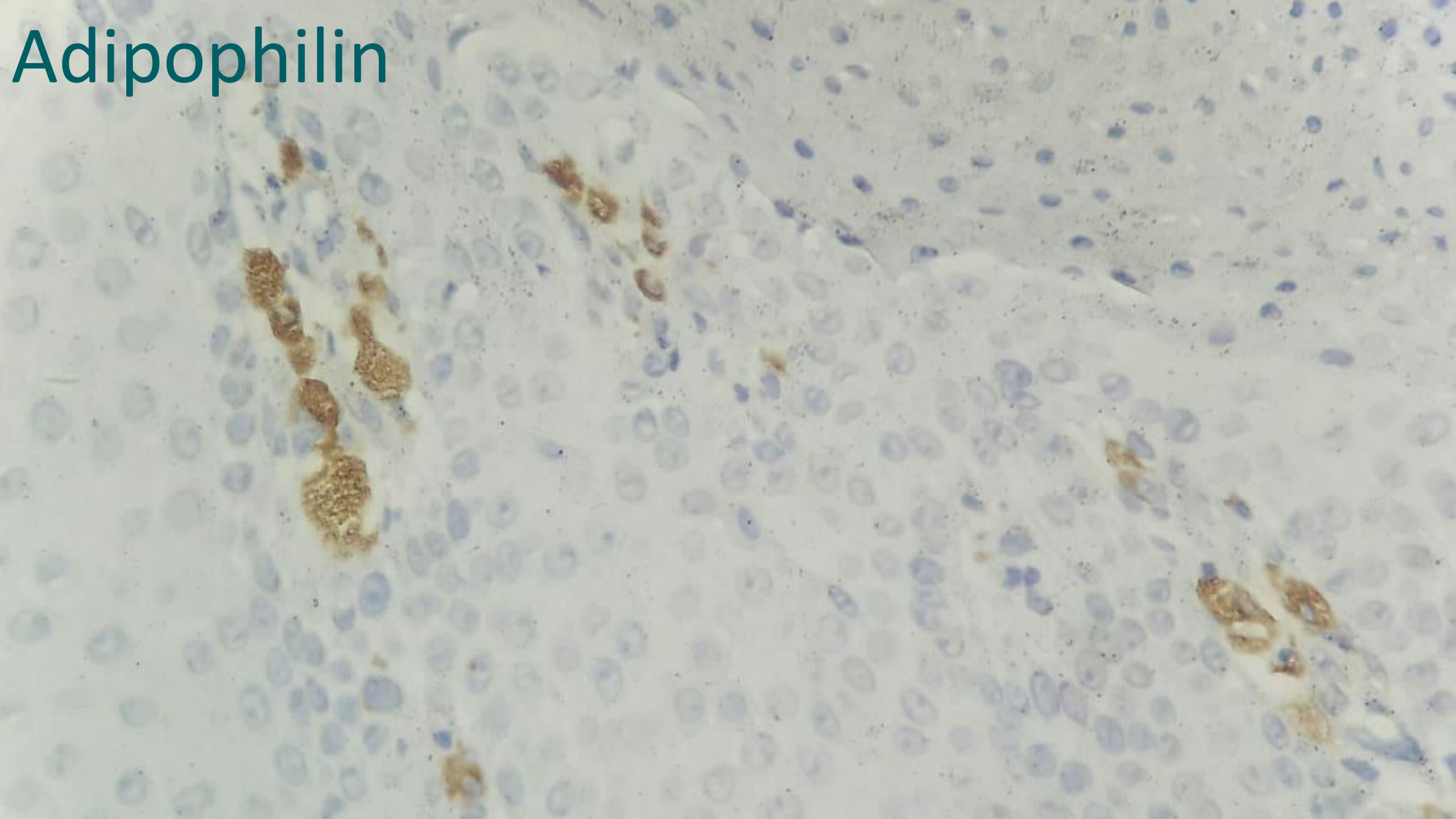
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Adipophilin



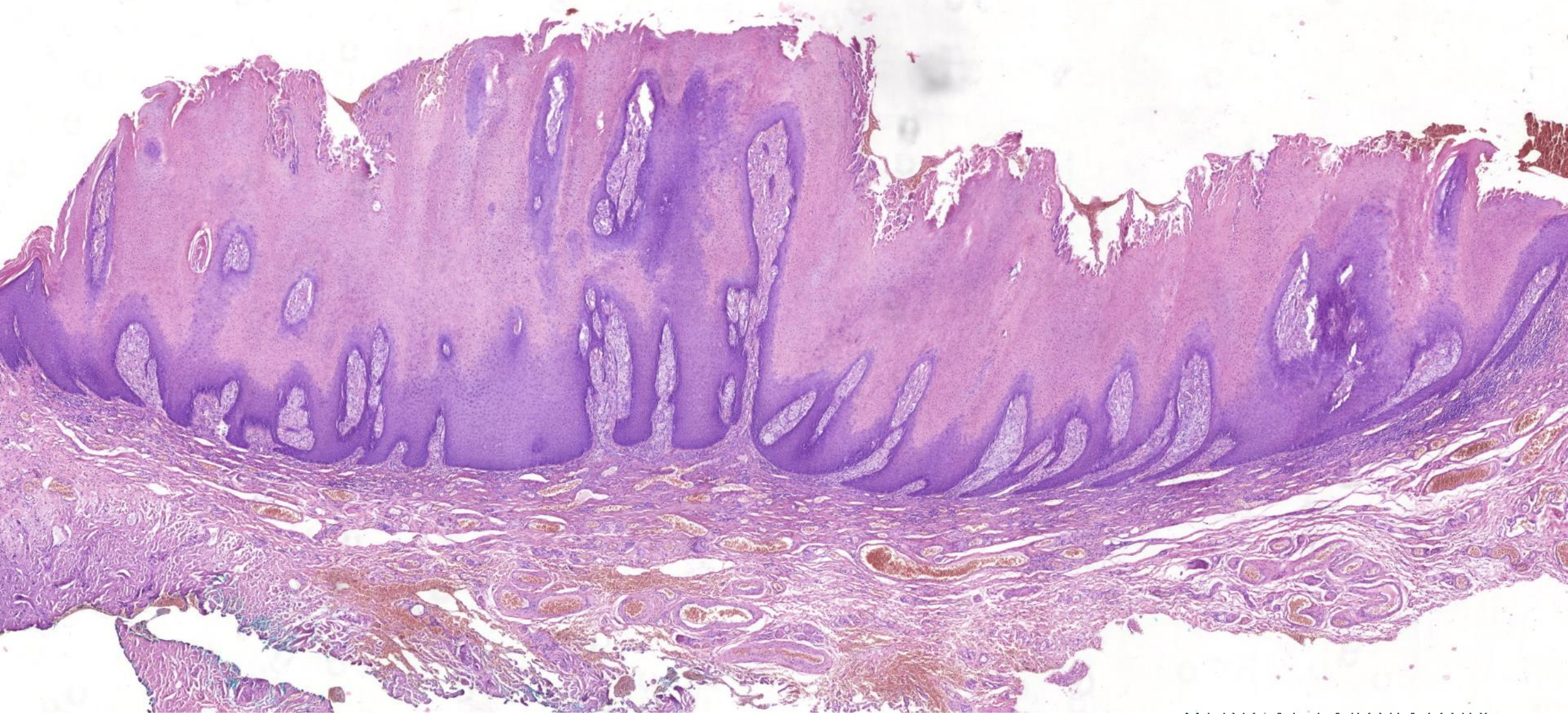




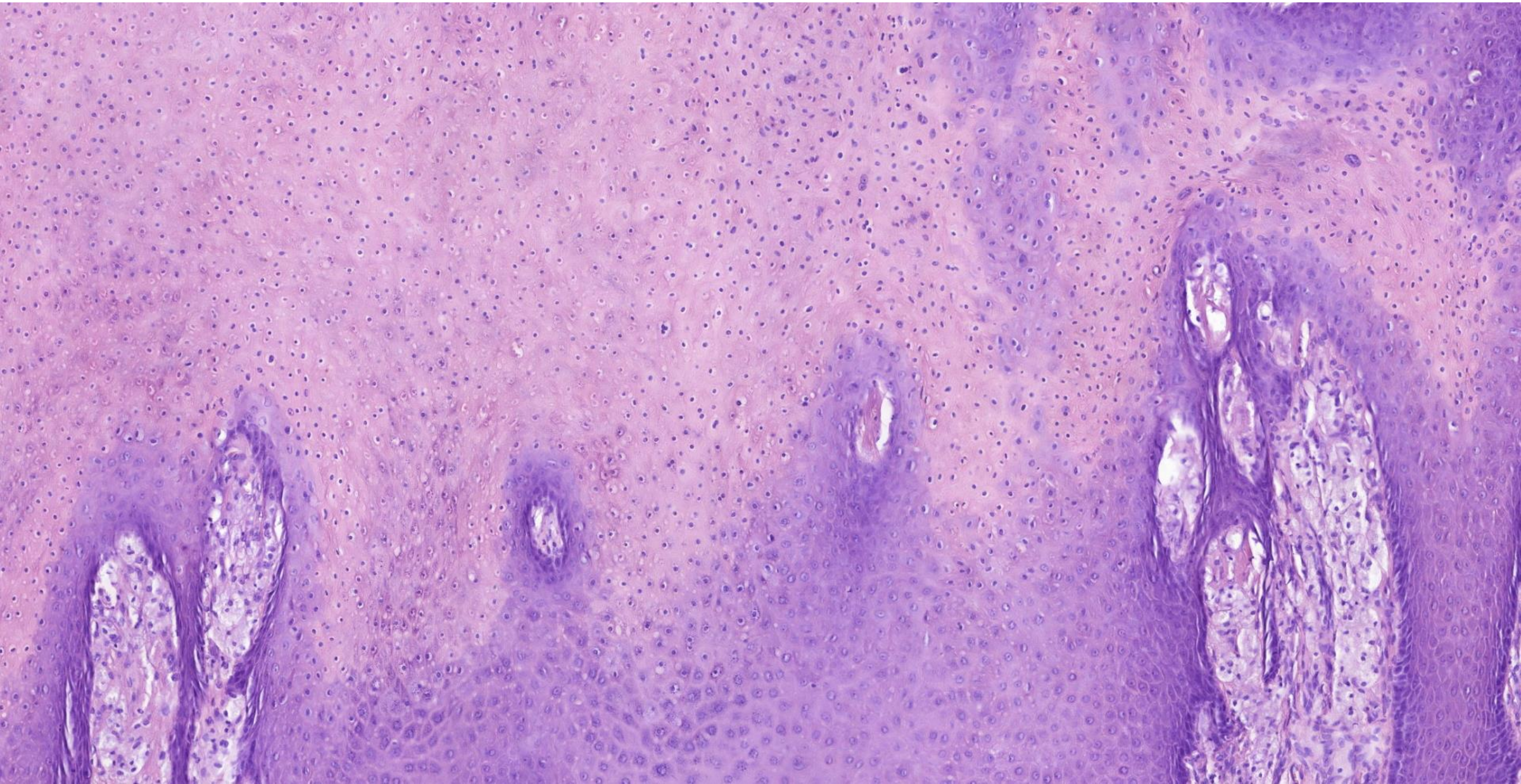
## Clinical case 4

- A 55-year-old female
- Admitted to the surgical department with solitary slightly itching lesion on the vulva
- Duration of the disease - 4 years with slowly progression of tumor growth.
- Clinical diagnosis at the time of admission: r\o SCC, VIN or other tumors.

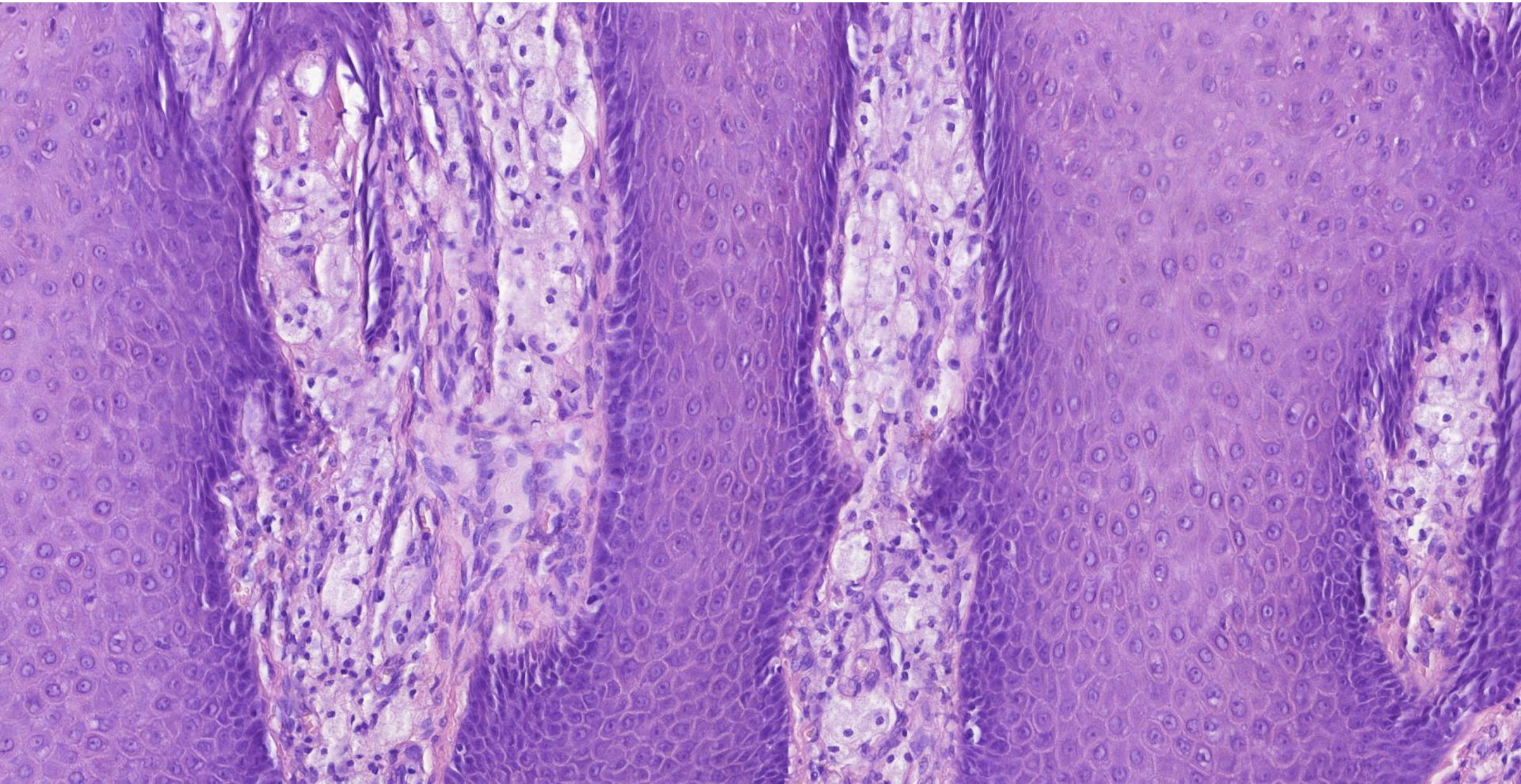
















## Your diagnosis?

- Irritated condyloma accuminatum
- Verruciform Genital-Associated Xanthoma or Vegas Xanthoma
- Verrucous carcinoma
- CHILD syndrome – associated Verruciform Xanthoma





## Your diagnosis?

- Irritated condyloma accuminatum
- **Verruciform Genital-Associated Xanthoma or Vegas Xanthoma**
- Verrucous carcinoma
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# Vegas (Verruciform Genital-Associated) Xanthoma: A Comprehensive Literature Review

Katherine M. Stiff · Philip R. Cohen

## ABSTRACT

**Introduction:** Verruciform xanthoma is a wart-like benign lesion. The classic histologic appearance consists of foamy histiocytes within elongated dermal papillae and epithelial acanthosis. The lesion most commonly occurs in the oral cavity, but has been reported in extra-oral sites such as the penis, scrotum, and vulva. The clinical and histologic characteristics of verruciform genital-associated (Vegas) xanthomas of the penis, scrotum, and vulva are reviewed.

**Methods:** PubMed was used to search the following term: verruciform xanthoma. The relevant papers were obtained and reviewed.

There were 164 in men and 29 in women. Similar to verruciform xanthomas of the oral mucosa, they presented as asymptomatic lesions, demonstrated foam cells in the dermal papillae, and were typically managed successfully with surgical excision.

**Conclusion:** Verruciform xanthoma is a benign lesion characterized by a wart-like growth that is most commonly seen in the oral mucosa. Verruciform xanthomas of the genital region have been coined Vegas xanthomas. Vegas xanthomas have been reported in association with a variety of diseases, as well as in healthy individuals. Biopsy is required for diagnosis, and complete surgical excision is typically curative.





## Conclusion

- Rare skin conditions happen and we should be aware of them
- Importance of clinical morphological correlation
- Stay in touch with your clinicians





Ďakujem za pozornosť!