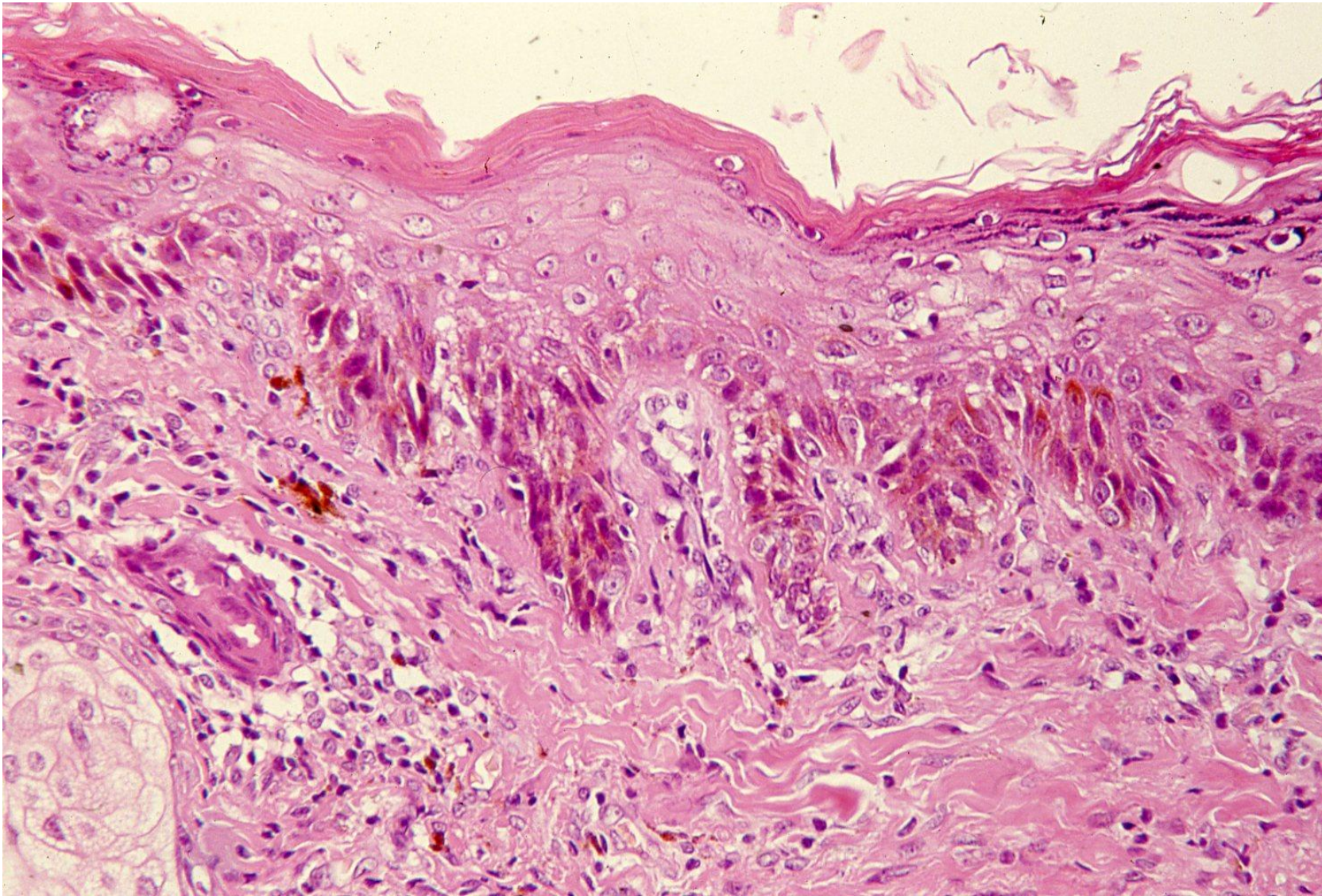


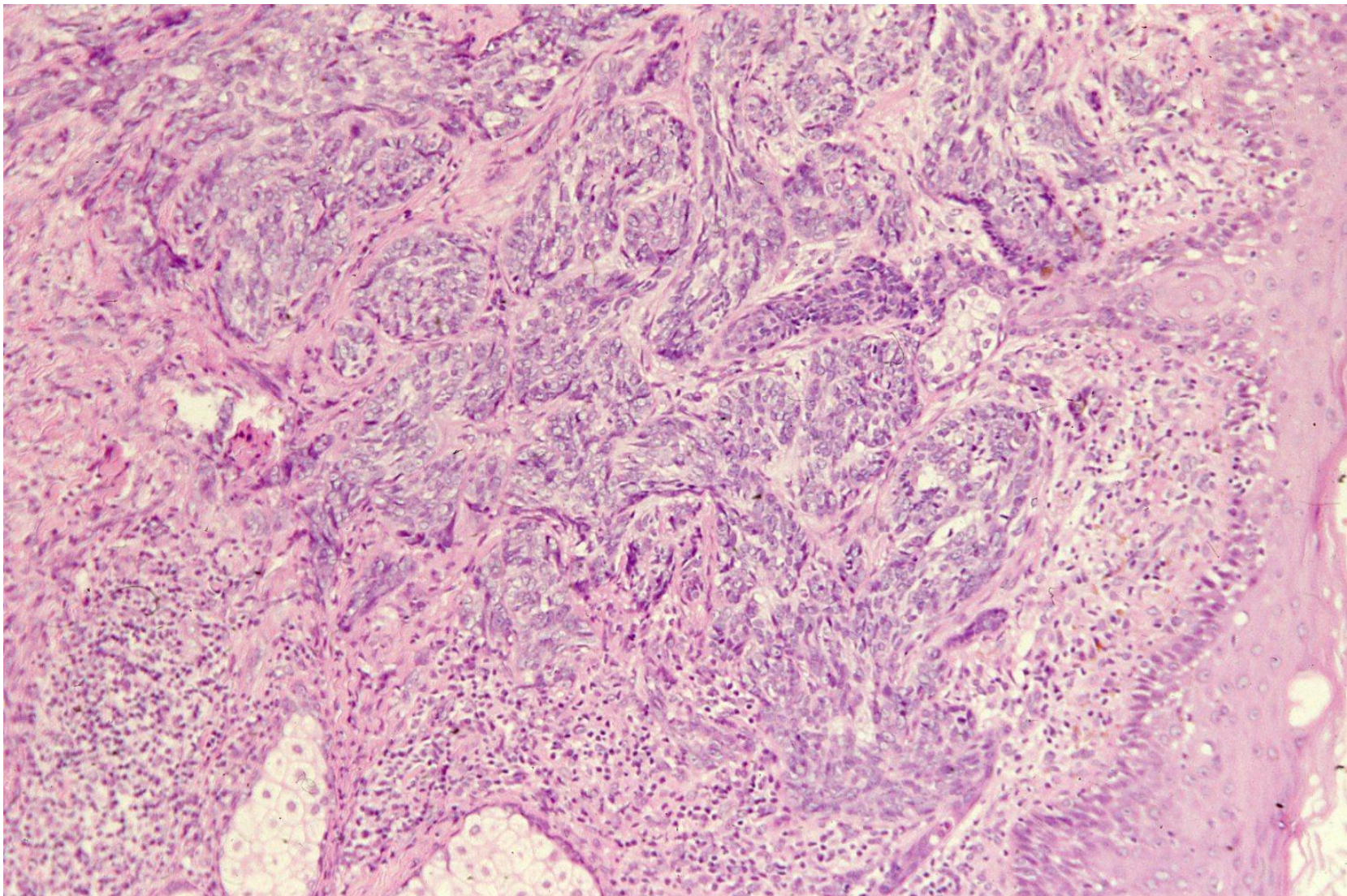
Xeroderma pigmentosum

Xeroderma pigmentosum (XP) is an autosomal recessive hereditary dermatosis caused by mutations in nucleotide excision repair. The nucleotide excision repair system is capable of removing ultraviolet-induced damage to DNA. XP demonstrates severe photosensitivity, skin pigmentation, development of malignant skin tumors, and occasionally progressive neurologic degeneration. There are subtypes of XP A-G and XP variants. XP progresses along with the accumulation of unrepaired DNA damage. The incidence of XP is relatively high in Japan. Non-melanoma skin cancer (basal cell carcinoma, keratoacanthoma and squamous cell carcinoma) commonly develops on the sun-exposed skin by the age of 9 years. Malignant melanoma may develop later by the age of 22. Squamous cell carcinomas of the anterior tongue may also be experienced. Ophthalmologic manifestations are as common as the skin manifestations, including photophobia, conjunctival injection, cataracts, conjunctivitis, blepharitis, pigmentation of the eyelid or conjunctiva, eye lid papilloma, ectropion, corneal vascularization, xerophthalmia and corneal scarring. The disease can be a model for photoaging and UV radiation-induced cancer.

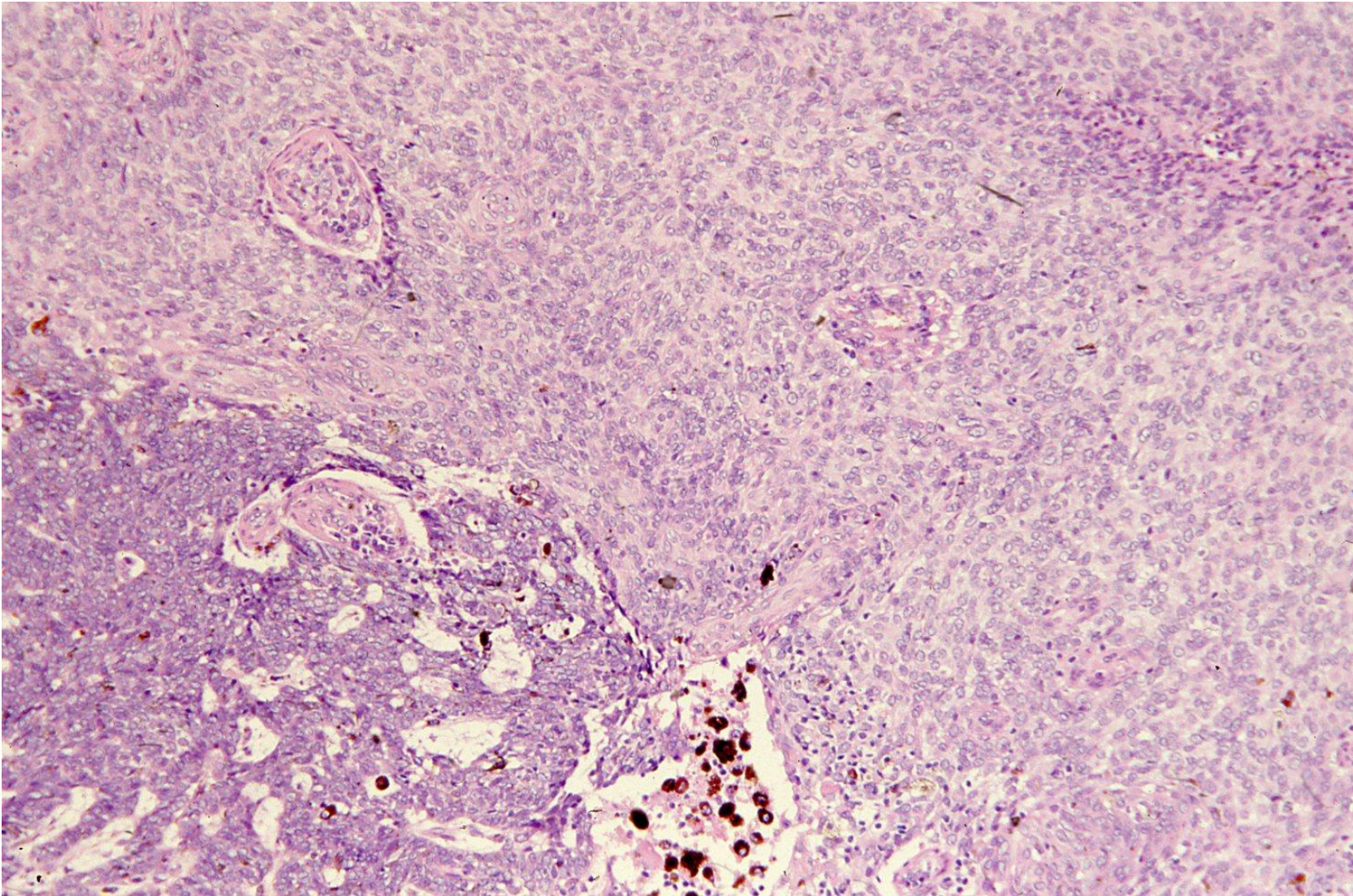
Ref.: Lucero R, Horowitz D. Xeroderma pigmentosum. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK551563/>



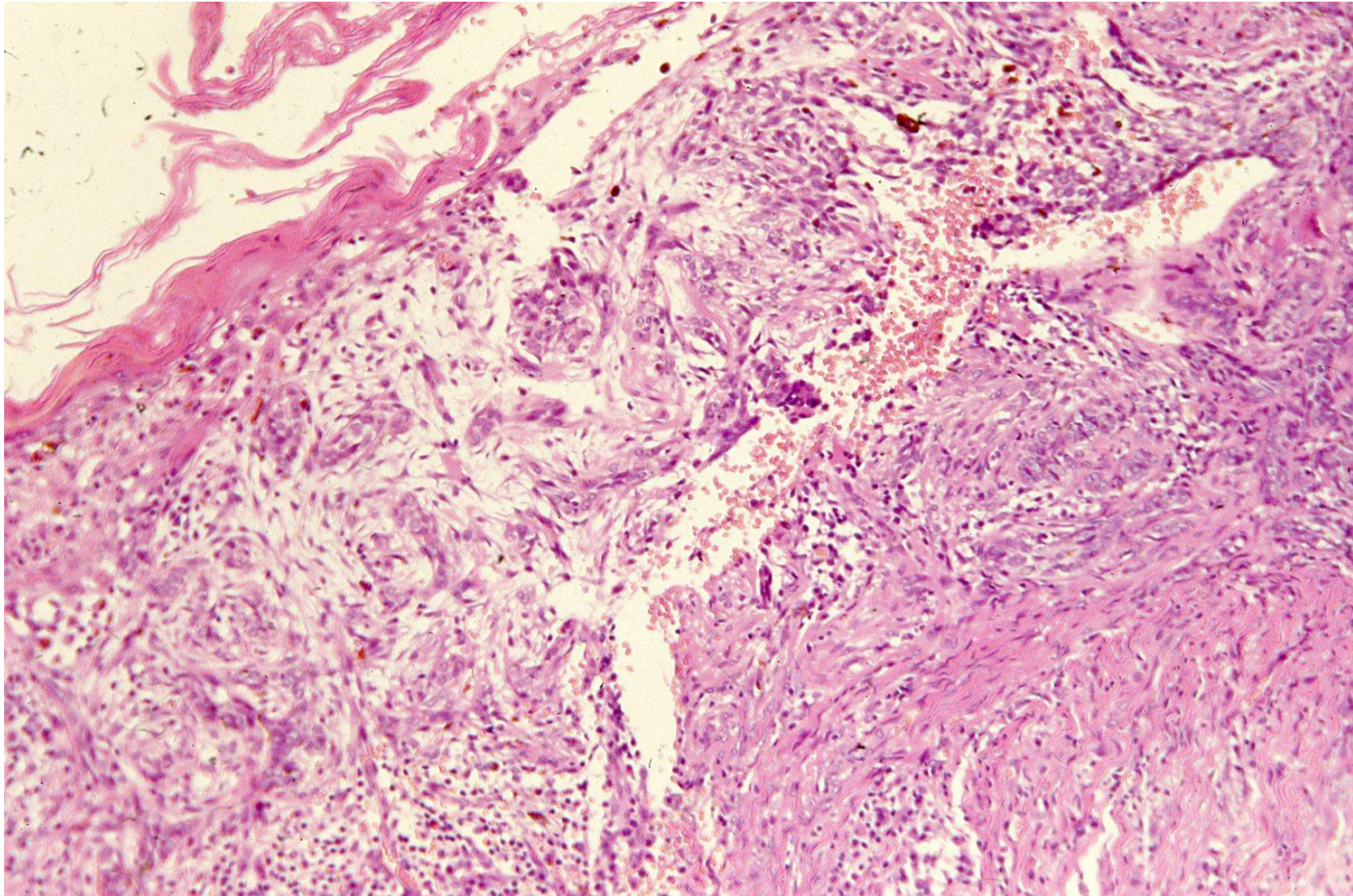
Xeroderma pigmentosum with accelerated basal cell activity, seen on the facial skin of a 19 y-o female patient (H&E-1).



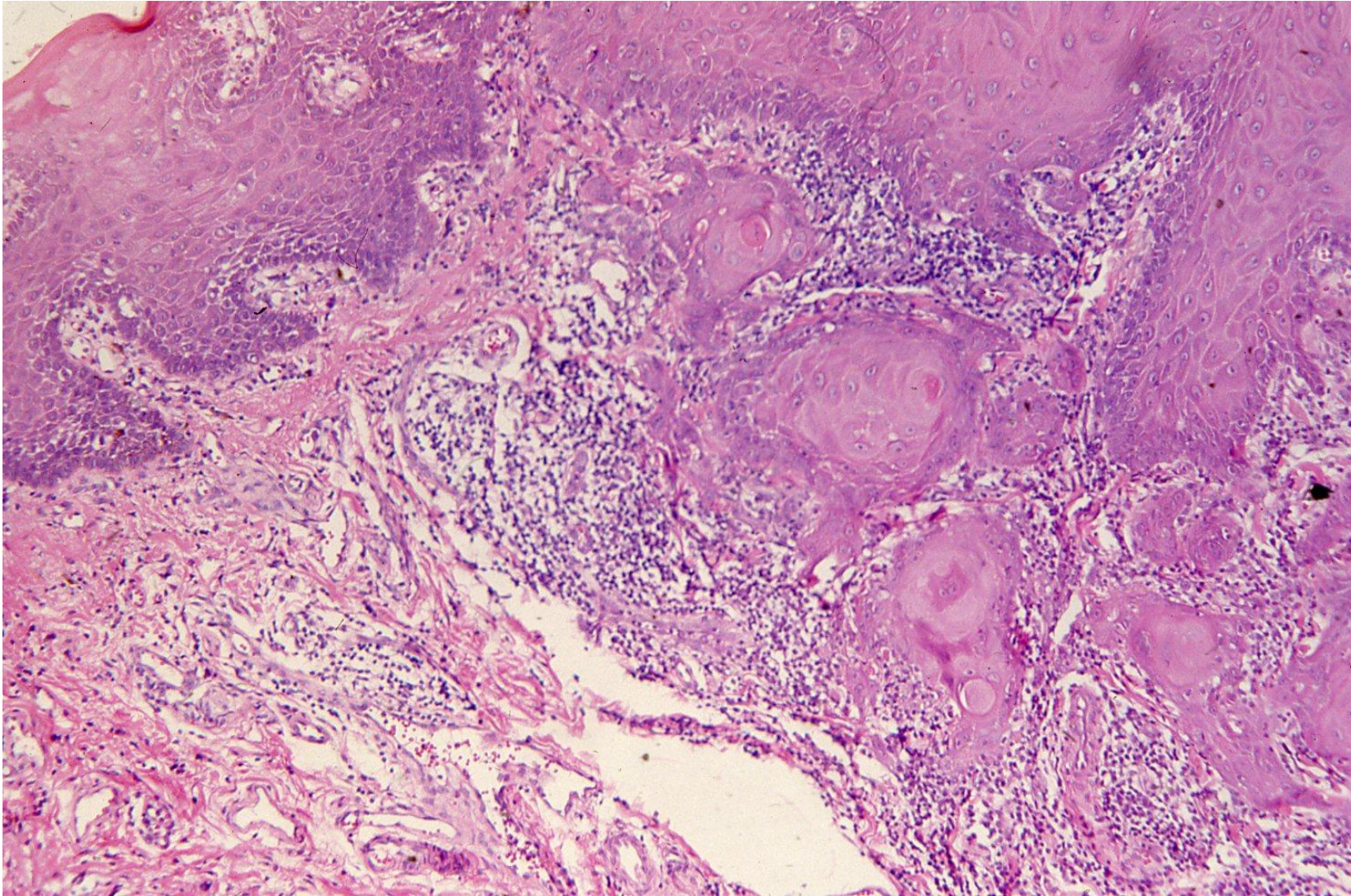
Xeroderma pigmentosum accompanying basal cell carcinoma, seen on the facial skin of a 19 y-o female patient (H&E-2).



Xeroderma pigmentosum accompanying basal cell carcinoma with squamous cell differentiation, seen on the facial skin of a 19 y-o female patient (H&E-3).



Xeroderma pigmentosum accompanying basal cell carcinoma with squamous cell differentiation, seen on the facial skin of a 19 y-o female patient (H&E-4).



Xeroderma pigmentosum accompanying invasive squamous cell carcinoma, seen on the facial skin of a 66 y-o female patient (H&E-5).