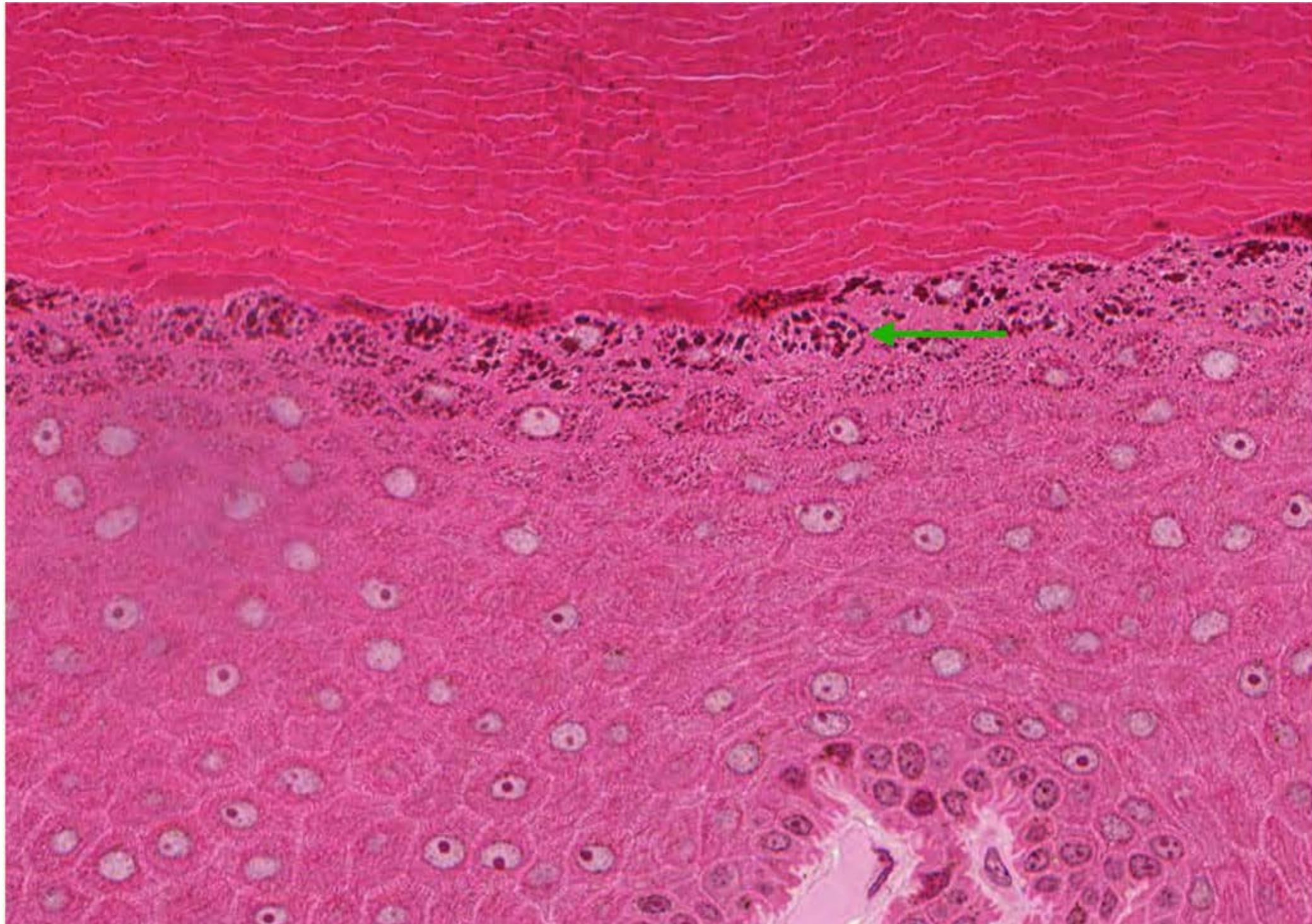


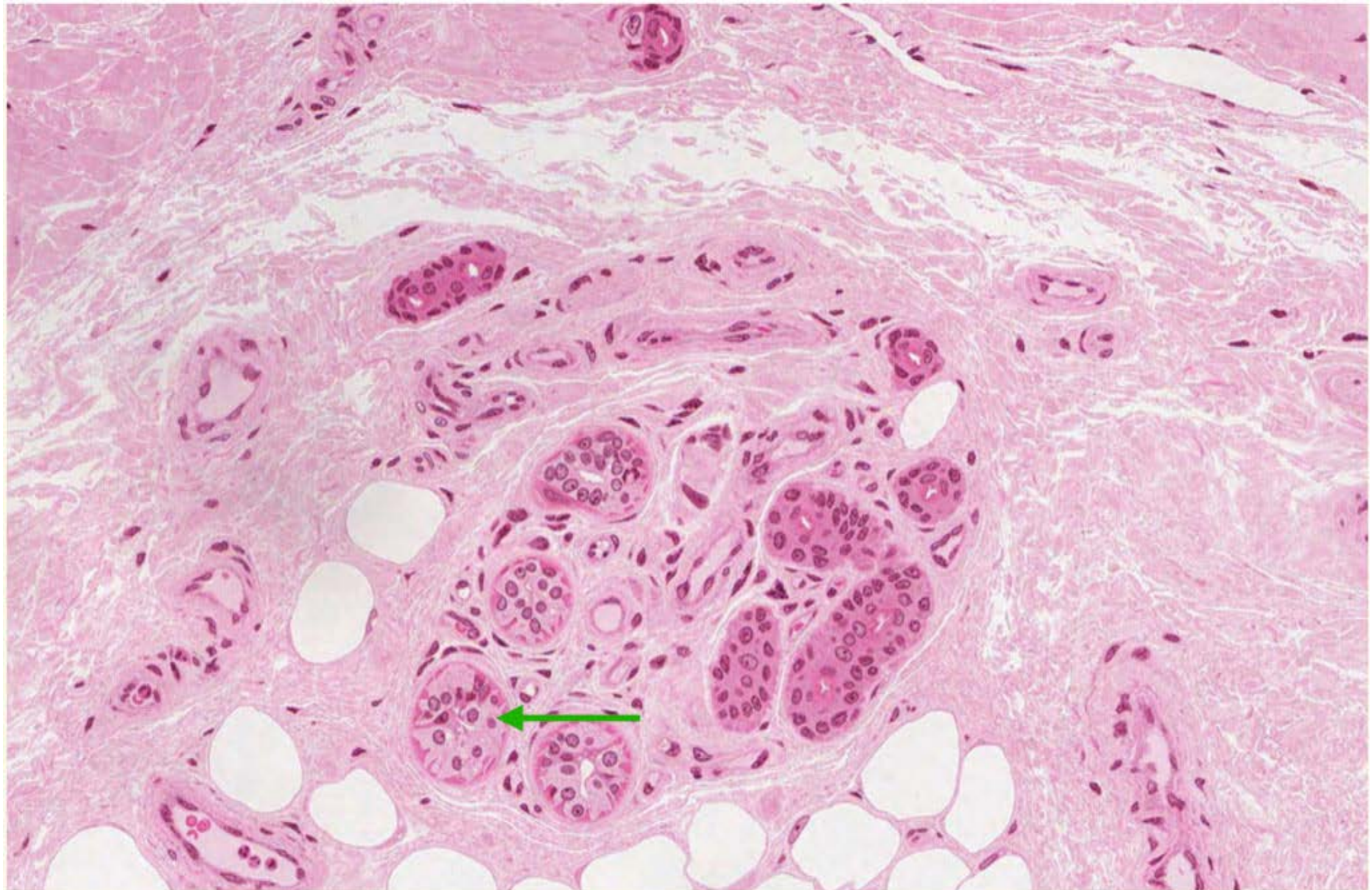
1. What is the function of the contents of the dark-staining granules in the cells in this region?

- Synthesize melanin
- Form lipid barrier
- Store serotonin
- Crosslink keratin filaments



2. Which best describes the fluid produced by these cells?

- Oily
- Isotonic
- Hypertonic
- Hypotonic



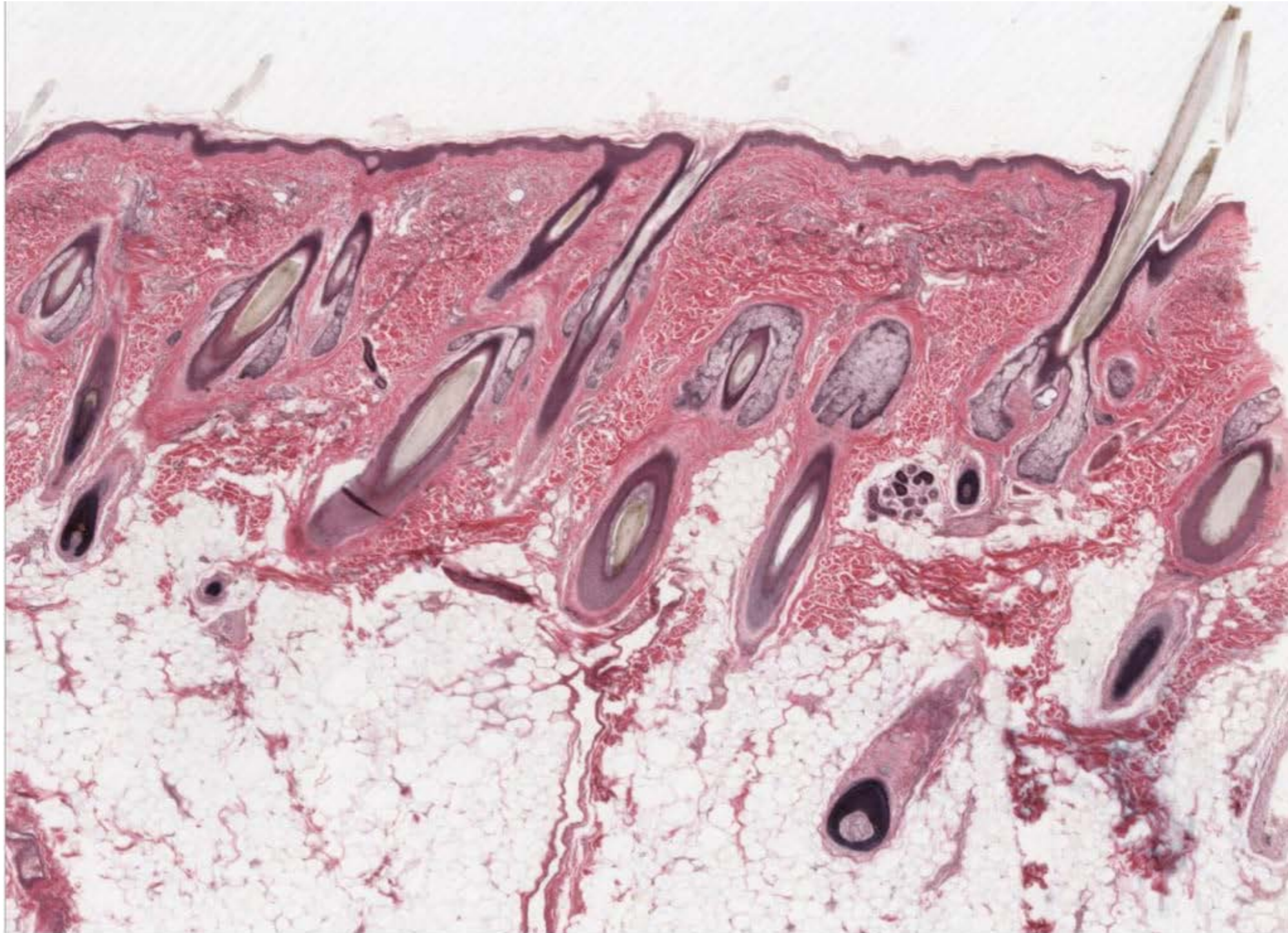
3. A mutation in a gene encoding which protein would most significantly compromise the mechanical integrity of this region?

- Integrin
- Connexin
- Cadherin
- Occludin



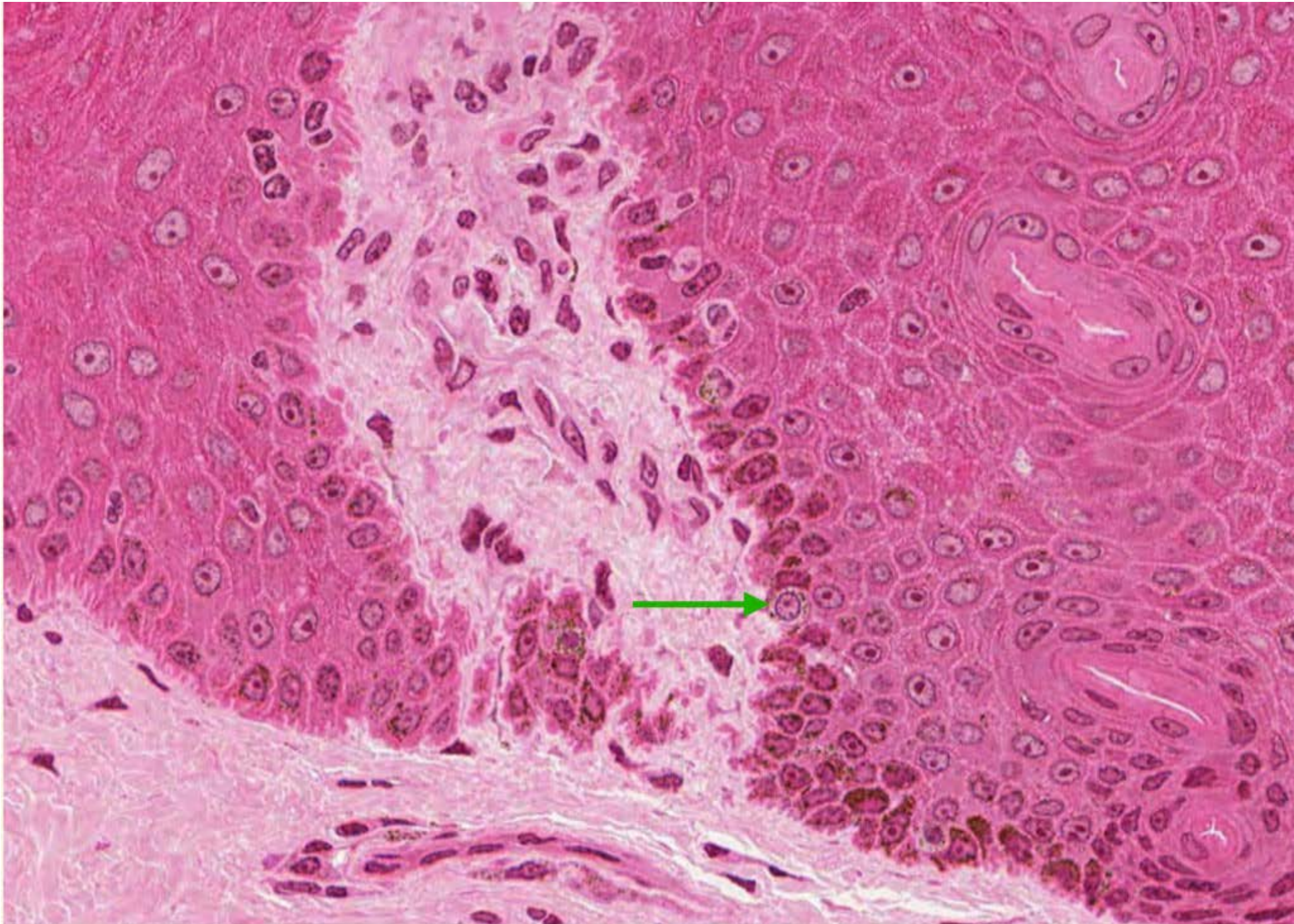
4. This section of skin was likely taken from which region of the body?

- Palms
- Finger tips
- Soles of feet
- Scalp

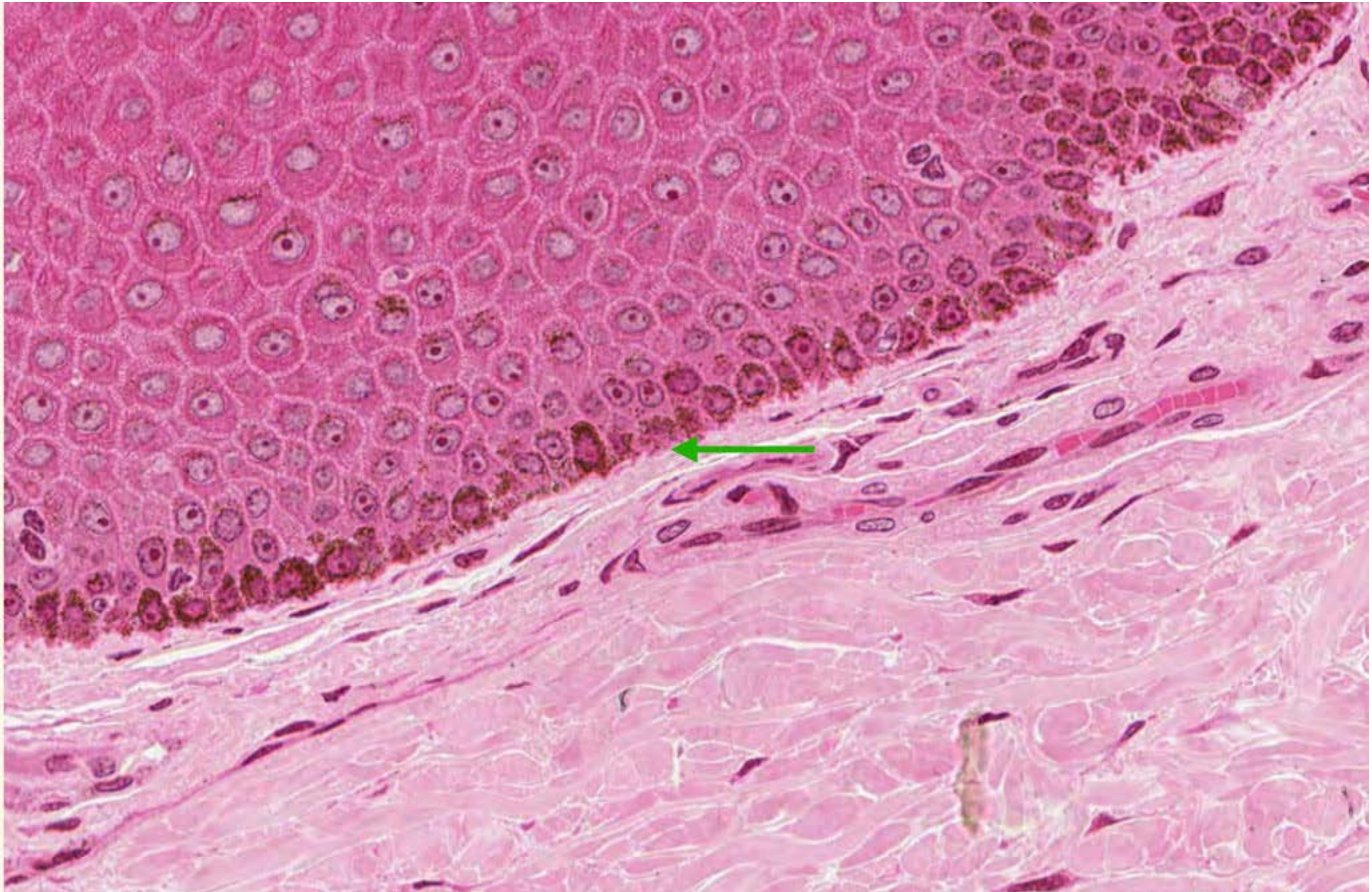


5. What is the primary function of the cell indicated by the arrow?

- Generate an immune response
- Generate new keratinocytes
- Synthesize cytokeratin
- Synthesize melanin

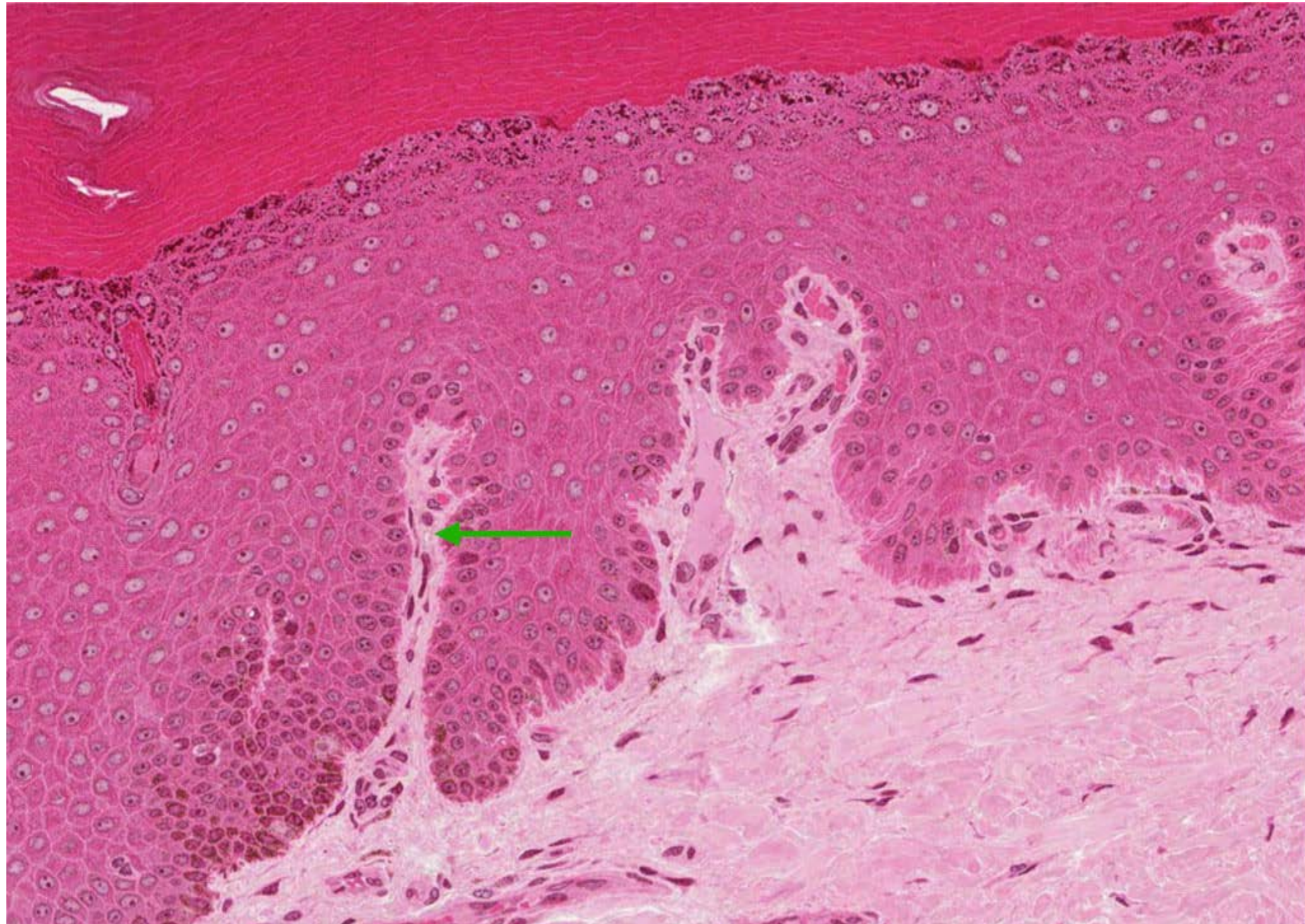


6. If you wanted to stain this region with an antibody for diagnostic purposes, an antibody protein would work best?
- Type I collagen
 - Laminin
 - Cadherin
 - Keratohyaline

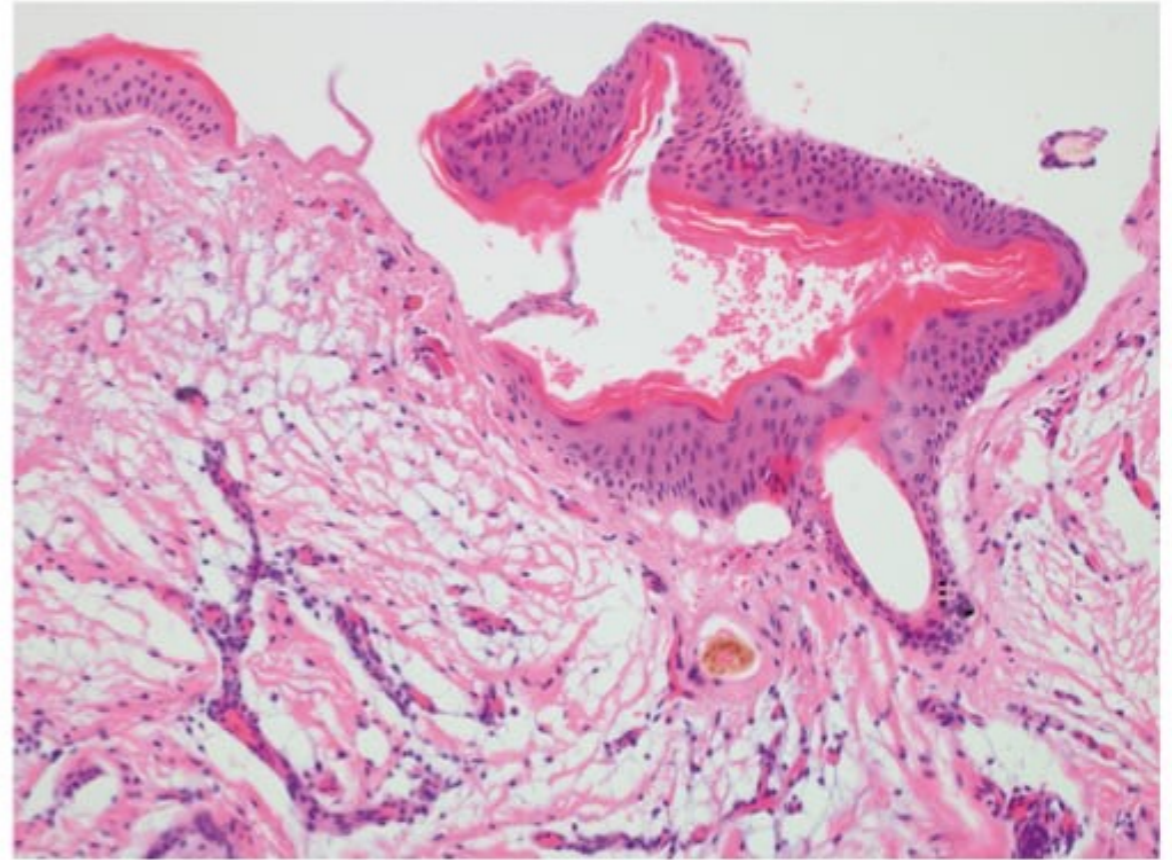


7. What best describes the function of this projection of the dermis?

- Promote more efficient delivery of nutrients to epidermis
- Promote delivery of sweat to surface of epidermis
- Promote adhesion with epidermis
- Promote turnover of epidermis

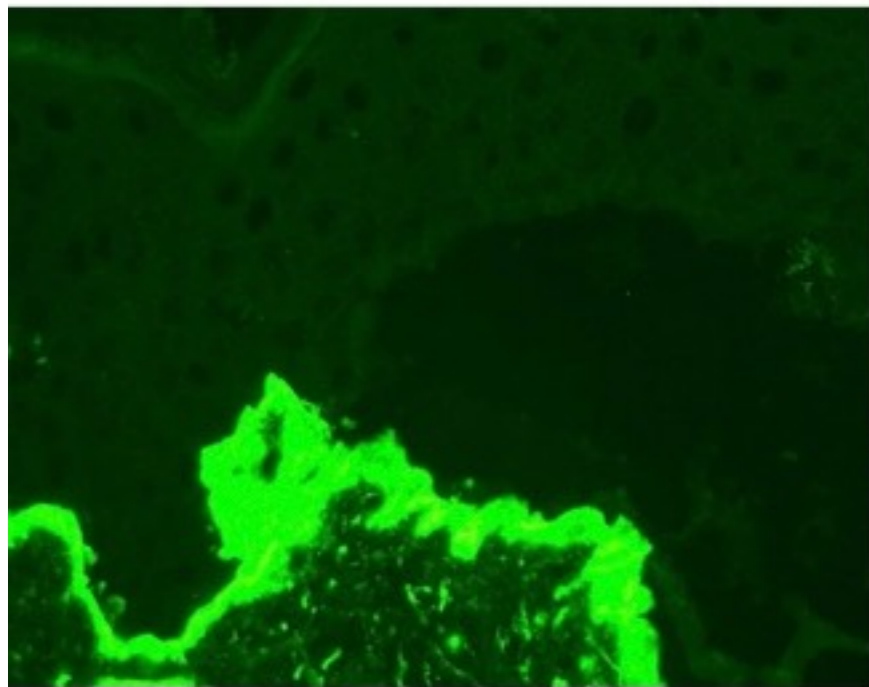


You are treating a child who develops skin blisters. A biopsy analyzed stained by H&E results in the image below. What information can you obtain from the image?

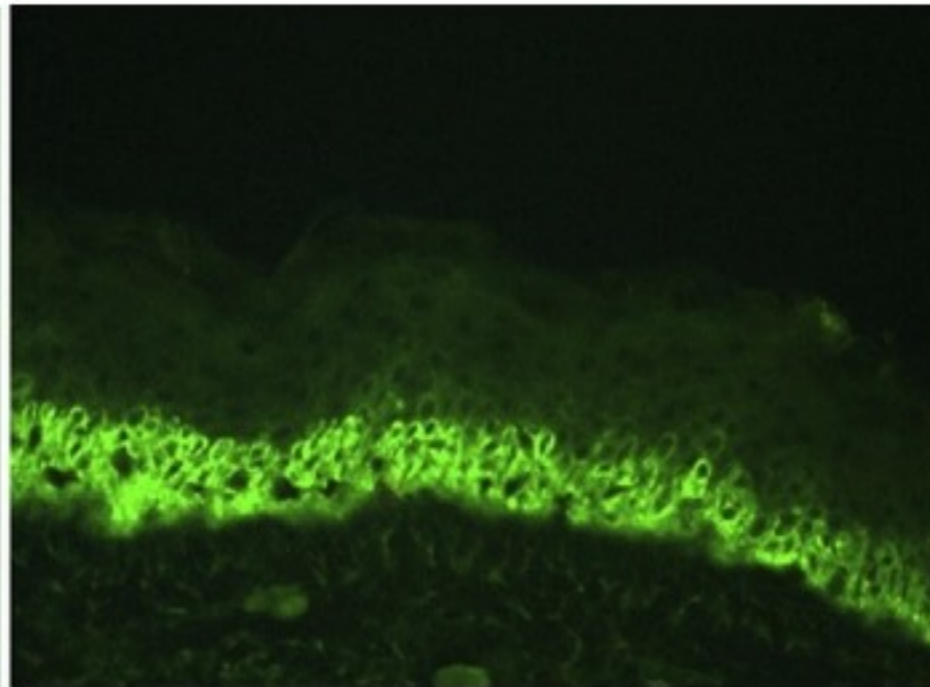


You send a sample of the biopsy to a lab where they will screen the biopsy with a set of antibodies against specific proteins in skin. Images from antibodies against three different proteins are shown below. What information is gained from the images? What pathology is suggested by the results?

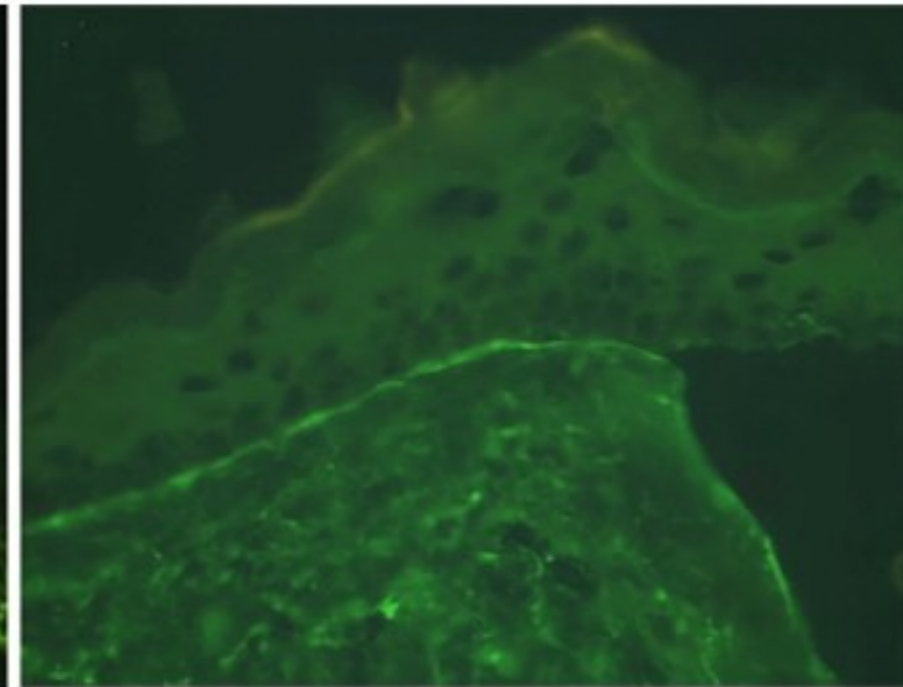
Type IV Collagen



Keratin 14



Laminin 322

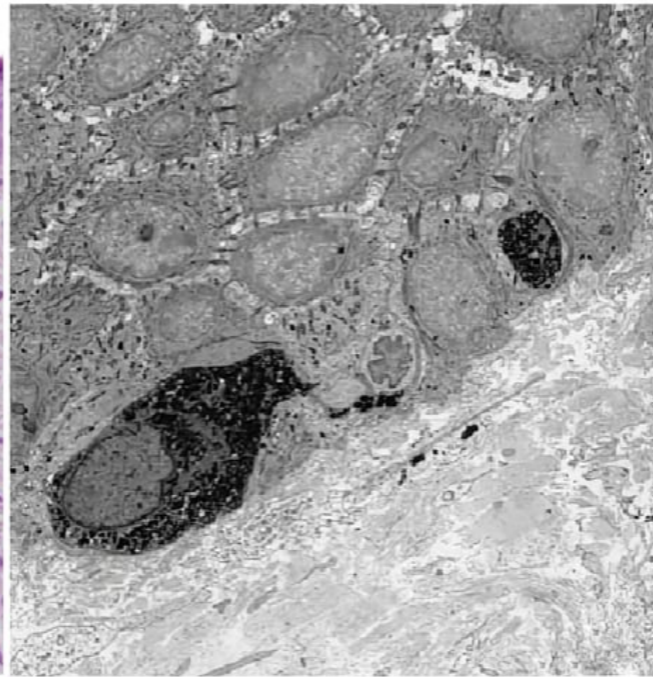
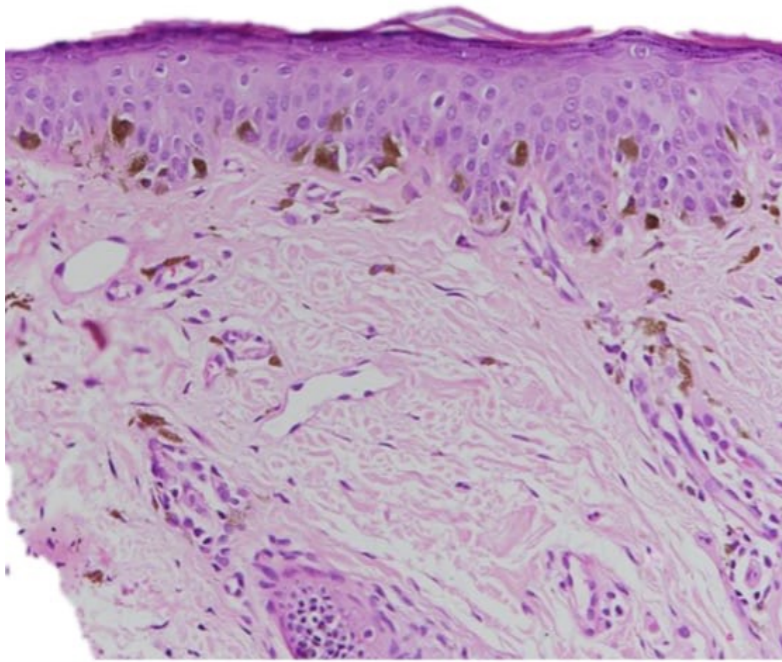


You also perform genome sequencing on a sample from the patient. How would mutations in coding and non-coding regions of the gene generate the staining pattern and blisters in the skin?

Are there non-genetic causes that could generate a similar phenotype?

Besides the pain of the blister, what other concerns would you have for the patient? Discuss potential treatment options. Do any provide long-term cures?

A 2-month old presents with hypotonia, silvery-greyish hair and very light colored skin.
A skin biopsy is processed for H&E and electron microscopy produces the images below.



A mutation in which gene most likely leads to the symptoms?

- Tyrosinase
- Integrin
- Myosin V
- Tyrosine Transporter