

Penile Cancer in a Man With Netherton Syndrome

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Netherton syndrome (NS) is a rare autosomal recessive skin disease with severe skin inflammation and scaling, a specific hair shaft defect (trichorrhexis invaginata or bamboo hair), and severe atopic manifestations including atopic dermatitis and hay fever with high serum immunoglobulin E levels and hypereosinophilia. NS is caused by loss-of-function mutations in serine protease inhibitor of Kazal-type 5 (*SPINK5*) encoding lympho-epithelial Kazal-type-related inhibitor (*LEKTI*) expressed in the stratified epithelia. We report the first case of penile squamous cell carcinoma in a patient with NS. *UROLOGY* 85: e21–e22, 2015. © 2015 Elsevier Inc.

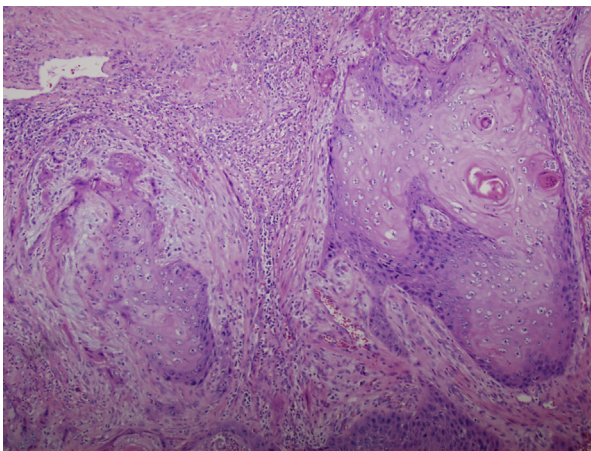


Figure 1. Well-differentiated squamous carcinoma infiltrates to level of corpora (hematoxylin and eosin stain, magnification $\times 100$).

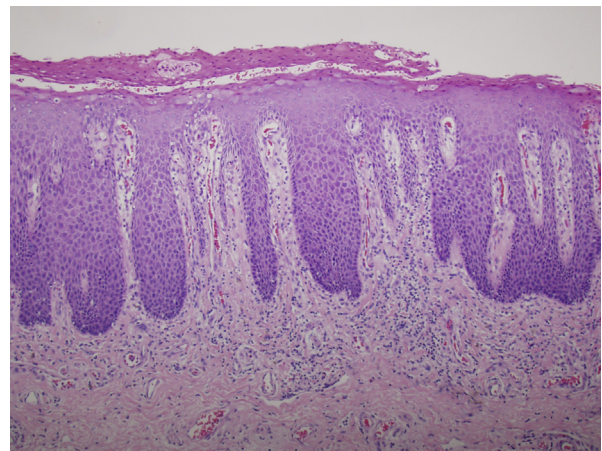


Figure 2. Photomicrograph of non-neoplastic skin shows psoriasiform hyperplasia, absent granular layer, parakeratosis, and focal detachment of stratum corneum. Note that the outermost nucleated layers do not flatten normally; inflammatory lymphoid infiltrate is seen in the upper dermis (hematoxylin and eosin stain, magnification $\times 100$).

Netherton syndrome (NS) is a rare and severe autosomal recessive genodermatosis caused by loss-of-function mutations in serine protease inhibitor of Kazal-type 5 (*SPINK5*) encoding lympho-epithelial Kazal-type-related inhibitor (*LEKTI*) expressed in the stratified epithelia.¹ Patients have impaired regulation of Th2 response to allergens and a defective response to skin bacterial infections.² There are 5 NS patients reported in the literature who developed squamous cell carcinoma (SCC).^{3–8}

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We report the first case of penile SCC in a 42-year-old man with NS. NS was diagnosed in childhood, and history was significant for phimosis. During dorsal slit for circumcision, he was found to have a fungating mass on the glans penis. Biopsy showed a well-differentiated SCC with lamina propria invasion. He underwent partial penectomy, and pathology showed a well-differentiated $3.8 \times 3.0 \times 2.5$ cm SCC invading corpora cavernosa and spongiosa (Fig. 1). This specimen was negative for human papillomavirus deoxyribonucleic acid. Figure 2 shows NS-associated non-neoplastic skin changes. At this time, on clinical examination, the patient had enlarged, nontender, fixed left inguinal lymph nodes. A positron emission tomography scan showed pathologically enlarged bilateral pelvic and inguinal lymph nodes with mild hypermetabolic activity (Fig. 3). The patient underwent bilateral inguinal and pelvic lymphadenectomy.

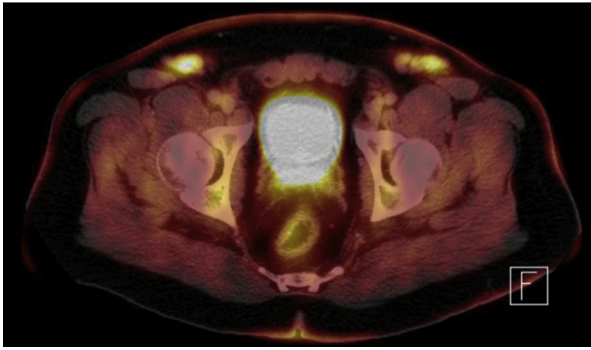


Figure 3. Axial positron emission tomography section showing bilateral enlarged inguinal nodes with mild hypermetabolic activity.

All lymph nodes showed dermatopathic lymphadenitis and no metastatic SCC. The patient is free of recurrence at 24 months of follow-up.

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