
DECEMBER IOTADERMA (#310)

Dermatology is filled with lots of words ending in "...us", such as pruritus, chronicus, planus, nitidus, etc. Do you happen to know what strophulus is?

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Answer will appear in the *JAAD* Online section of the January issue of the Journal.

NOVEMBER IOTADERMA (#309)

Question: All of us have done things in practice and at home on a whim. Do you happen to know what the **WHIM** syndrome is, and what its easily observable and common dermatologic finding is?

Answer: The **WHIM** syndrome is an exceedingly rare genetic condition with several cardinal features: a persistent neutropenia, early onset intermittent severe and diverse infections (due to an associated B cell lymphopenia and hypogammaglobulinemia), and myelokathexis of the bone marrow with apoptosis, slow neutrophil release, and myeloid hyperplasia.¹⁻³ Most importantly for dermatologists, however, is the **W** part of the acronym: affected patients have a markedly enhanced propensity for infections by the human papilloma virus (HPV), leading to the finding of many ordinary type warts (in 61% of patients), as well as the presence of condylomata acuminata and various HPV induced skin cancers (in 16% of patients).³ There appear to be several genetic defects which produce **WHIM**, especially the heterozygous C-terminus deletional mutations of parts of the intracellular carboxy terminus of the chemokine receptor CXCR4 leading to a gain in function, as well as a characteristic hyperresponsiveness of leukocytes to SCF1 (the known ligand of CXCR4) which promotes defective GRK3 phosphorylation mediated downregulation of CXCR3.^{1,2} Treatments include immunoglobulin and granulocyte stimulating factor infusions.³

REFERENCES

1. Toshinao K, Malech HL. WHIM Syndrome: Congenital Immune Deficiency Disease. *Curr Opin Hematol*. 2009;16(1):20-26.
2. Naoum FA. A report of WHIM syndrome (myelokathexis) - clinical features and bone marrow morphology. *Rev Bras Hematol Hemoter*. 2011;33(5):393-394.
3. Dotta L, Notarangelo LD, Moratto D, et al. Long term outcome of WHIM syndrome in 18 patients: high risk of lung disease and HPV-related malignancies. *J Allergy Clin Immunol Pract*. 2019;7(5):1568-1577.

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