Question 32 – Week of February 7

This syndrome is associated with pancreatic exocrine insufficiency, nasal alar hypoplasia, teeth abnormalities, short stature, psychomotor retardation, congenital deafness, and ectodermal scalp defects.

A. Shwachman-Diamond Syndrome
B. Turcot's Syndrome
C. Muir-Torre Syndrome
D. Johanson-Blizzard Syndrome

Answer: D

Johanson-Blizzard Syndrome is an autosomal recessive disorder with the above noted features. The gene defect appears to be associated with ubiquination of proteins. Shwachman-Diamond Syndrome results from a gene defect near the centromere of chromosome 7 and involves the Shwachman-Bodian-Diamond Syndrome gene (SBDS). It is autosomal recessive and characterized by pancreatic exocrine insufficiency, cyclic neutropenia, metaphyseal dysostosis, and growth retardation. Turcot's Syndrome is an autosomal dominant variant of HNPCC caused by mutations of one of the MMR genes and characterized by brain tumors and adenomatous oligopolyposis. Muir-Torre Syndrome is also an autosomal dominant variant of HNPCC, characterized by sebaceous adeomas, sebaceous epitheliomas, or sebaceous carcinomas and an internal malignancy, usually CRC, found in about half of patients.