

Question 27 – Week of January 3

Which of the following is an autosomal recessive disorder characterized by pancreatic exocrine insufficiency, cyclic neutropenia, metaphyseal dysostosis, and growth retardation?

- A. Gorlin Syndrome
- B. Bannayan-Ruvalcaba-Riley Syndrome
- C. Shwachman-Diamond Syndrome
- D. Cowden's Syndrome

Answer: C

Shwachman-Diamond Syndrome results from a gene defect near the centromere of chromosome 7 and involves the Shwachman-Bodian-Diamond Syndrome gene (SBDS). The above noted clinical syndrome is seen. Gorlin Syndrome is caused by mutations of the PTCH gene on chromosome 9q and is characterized by multiple nevoid basal carcinomas, skeletal abnormalities, odontogenic keratinocytes, macrocephaly, intracranial calcification, and craniofacial abnormalities. Infrequently these patients have juvenile polyps of the GI tract. Bannayan-Ruvalcaba-Riley Syndrome is an ill-defined, possibly autosomal dominant syndrome which could be a variant of juvenile polyposis with patients having developmental delay, macrocephaly, pigmented macules of the shaft and glans of the penis, and lipid storage myopathy. 50-60% of these individuals have germline mutations in the PTEN gene on chromosome 10q. Cowden's Syndrome is an autosomal recessive disorder with variable hamartomatous involvement of the skin, mucous membranes, GI tract, thyroid, and breast is associated with multiple facial trichilemmomas, verrucous skin lesions of the face and limbs, and cobblestone-like papules of the gingiva, buccal mucosa, and tongue.