Question 36 – Week of April 15

Mutation of which of the following genes results in autosomal dominant hereditary hemochromatosis?

A. HFE
B. Hemojuvelin
C. Ceruloplasmin
D. Transferrin receptor 2
E. Ferroportin

Answer: E

A mutation in a gene encoding for ferroportin (SLC40A1) is associated with an autosomal dominant form of hemochromatosis in unrelated European and Australian families. This gene encodes for a basolateral membrane protein that has a role in the movement of iron across the enterocytes and macrophages into the circulation. The other answers are all associated with autosomal recessive forms of hemochromatosis

Reference:

Njajou OT, et al. A mutation in SLC11A3 is associated with autosomal dominant hemochromatosis. Nat Genet. 2001;28(3):213.