Sphingolipidoses

TRUE/FALSE Questions

1. In normal subjects, hexosaminidase A has an alpha subunit but no beta subunit
2. Sandhoff's disease is an example of a beta subunit gene defect
3. The action of the hexosaminidases results in the release of N-acetyl-galactosamine from either ganglioside GM2 or globoside
4. In Gaucher disease, galactocerebrosides accumulate in macrophages
5. The bone lesion in Gaucher disease has been described as a *flask deformity*
6. A cherry red spot is a finding in GM1 and GM2 gangliosidosis as well as in one third of the patients with the infantile form of Niemann-Pick disease
7. Sandhoff observed that there was no ethnic tendency in the patients with a history similar to that seen in Tay-Sachs disease. Also, there was storage of globoside in visceral organs
8. In infantile Niemann-Pick disease the physical exam gives an important clue to the diagnosis

MULTIPLE CHOICE - SINGLE BEST ANSWER

1. Which one of the following metabolites is involved with ceramide in the biosynthesis of sphingomyelin?
   A. Surfactant
   B. Phosphatidyl serine
   C. UDP-Galactose
   D. UDP-glucose
   E. Phosphatidyl choline

2. Identify from the following options the single best answer related to fundamental aspects of Tay-Sachs and Sandhoff's disease

   A. In Sandhoff Disease, both hexosaminidase A and hexosaminidase B activities are defective
   B. The action of the hexosaminidases results in the release of N-acetyl-galactosamine from either ganglioside GM2 or globoside
   C. A carrier for Tay-Sachs Disease has normal total hexosaminidase activity
   D. Options A, B, and C are all correct statements

ANSWERS: F, T, T, F, T, T, T, T #1. E #2. D