CONTINUING MEDICAL EDUCATION EXAMINATION

Genetic Defects in B-Cell Development and Their Clinical Consequences

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CME Items

1. Your patient is a 4-year-old boy with recurrent otitis media and sinusitis. He has just been clinically diagnosed with common variable immunodeficiency. Which one of the following can you check to confirm the diagnosis?
   a) *LRBA*
   b) *TACI*
   c) *BAFF-R*
   d) *MSH5*

2. A 6-year-old boy presented with conical teeth and sparse hair and 2 episodes of pneumonia in 1 year. Which of the following are not probable in this case?
   a) Increased level of serum IgM
   b) Susceptibility to fungal infection
   c) History of delayed umbilical detachment
   d) Defect in *IKBKG* with X-linked inheritance

3. A consanguineous couple with a 6-month-old daughter diagnosed with agammaglobulinemia and CD19 <1% are coming for family planning and genetic counseling before their next pregnancy. Which of the following warrants further analysis?
   a) CD40 deficiency
   b) CD81 deficiency
   c) UNG deficiency
   d) Igα deficiency

4. All the genes involved in hyper-IgM syndrome affect both class-switching defect and somatic hypermutation, except:
   a) *NEMO*
   b) *UNG*
   c) *AID*
   d) *CD40L*

5. A high-resolution computed tomography scan was performed in a 4-year-old girl with pneumonia. She was diagnosed with *Pneumocystis jiroveci* pneumonia. Her immunologic profile reveled the following results: IgG, 302 mg/dL; IgM, 132 mg/dL; IgA, 9 mg/dL; CD19, 6.6%. Which gene is more compatible with these manifestations?
   a) *AID*
   b) *CD40*
   c) *UNG*
   d) *PMS2*

6. In a male patient with a common variable immunodeficiency phenotype (IgG, 60 mg/dL; IgA, 5 mg/dL; IgM, 20 mg/dL; CD19, 10%) and non-consanguineous parents, which of the following disorders should be included in the differential diagnosis?
   a) X-linked agammaglobulinemia (*BTK* gene)
   b) X-linked hyper-IgM syndrome (*CD40L* gene)
   c) X-linked lymphoproliferative disease (*SH2D* gene)
   d) AID deficiency with a defect in the C terminus

7. An adult patient with terminal B-cell deficiency and lymphopenia, reduced plasma cell counts, and antitetanus antibody defect had a *BAFF-R* polymorphism. Which condition is more probable in the humoral immune profile of this case?
   a) Normal IgA level
   b) Elevated IgM level
   c) Normal IgG level
   d) Elevated IgE level

8. Which of the following proteins should be joined to λ5 for homology of the J region and BCR signaling?
   a) BLNK
   b) Igα
   c) PIK3R
   d) VpreB

9. Which of the following clinical phenotypes is compatible with a human patient with PIK3R deficiency?
   a) Idiopathic thrombocytopenic purpura, lymphopenia, and hypogammaglobulinemia
   b) Colitis, neutropenia, and agammaglobulinemia
   c) Juvenile rheumatoid arthritis, enteropathy, and hypogammaglobulinemia
   d) Gastrointestinal cancer, CD19 <1%, and agammaglobulinemia

10. A significant group of patients with common variable immunodeficiency had a defect in a protein involved in the CD19 complex, which mediates regulation of B-cell development. Which item is not one of the components of this complex?
    a) CD81
    b) CD28
    c) CD21
    d) CD225