

A 4-month-old boy is admitted to the intensive care unit for respiratory distress due to pneumonia. The infant developed cough and tachypnea 2 days ago with progressive worsening of hypoxia and work of breathing. He has had 2 months of poor weight gain and diarrhea, which have not improved despite changing to a hydrolyzed formula. The infant lives with his foster parents, who have 2 older children and a dog. The biological mother did not receive prenatal care after the first trimester. The child's birth weight was at the 60th percentile and current weight is at the 3rd percentile. Temperature is 39.5 C (103.1 F) and pulse oximetry is 74% on room air. Physical examination shows extensive oral thrush and generalized lymphadenopathy. Auscultation of the heart and lungs reveals a soft systolic ejection murmur and faint bilateral crackles. The remainder of the examination is normal. Laboratory results are as follows:

Hematocrit	38%
Platelets	260,000/mm ³
Leukocytes	18,000/mm ³
Neutrophils	50%
Lymphocytes	45%

Endotracheal tube aspirate reveals *Pneumocystis jirovecii*. Which of the following abnormalities is most likely present in this patient?

- ☐ A. Abnormal chloride ion transporter
- ☐ B. Decreased B lymphocyte count
- ☐ C. Decreased CD4+ lymphocyte count
- ☐ D. Defect in dynein
- ☐ E. Deficiency of adenosine deaminase
- ☐ F. Impaired phagocytosis

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Endotracheal tube aspirate reveals *Pneumocystis jirovecii*. Which of the following abnormalities is most likely present in this patient?

- ☐ A. Abnormal chloride ion transporter [7%]
- ☐ B. Decreased B lymphocyte count [3%]
- ☒ C. Decreased CD4+ lymphocyte count [69%]
- ☐ D. Defect in dynein [1%]
- ☐ E. Deficiency of adenosine deaminase [16%]
- ☐ F. Impaired phagocytosis [3%]

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Explanation:

User Id: [REDACTED]

HIV in infancy

HIV in infancy	
Risk factors	<ul style="list-style-type: none"> • High maternal viral load • Breastfeeding by infected mother
Clinical features	<ul style="list-style-type: none"> • Failure to thrive • Chronic diarrhea • Lymphadenopathy • <i>Pneumocystis pneumonia</i>
Diagnosis	<ul style="list-style-type: none"> • DNA polymerase chain reaction testing • Persistence of HIV antibody after age 18 months
Treatment	<ul style="list-style-type: none"> • Immediate combination antiretroviral therapy

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This infant's **chronic diarrhea, lymphadenopathy, failure to thrive**, and severe infections are suggestive of an immunodeficiency syndrome. Specifically, his history of infection with **opportunistic organisms** (eg, *Pneumocystis*, *Candida*) suggests a T lymphocyte deficiency. **HIV infection** presents with a normal absolute lymphocyte count and **CD4+ T lymphocyte deficiency** as most HIV replication occurs in CD4+ T cells. Because maternal antibodies may be present in HIV-negative children age <18 months, DNA PCR testing of blood can confirm the diagnosis. After age 18 months, persistence of HIV antibody is confirmatory of infection.

Infants can acquire HIV infection via the transplacental route or, more commonly, during delivery. High maternal viral load is the most important risk factor in transmission. Prenatal screening and maternal antiretroviral therapy have markedly reduced infection rates in the United States, but infections still occur when mothers are infected late in pregnancy or have insufficient prenatal care.

(Choice A) Abnormal chloride ion transport occurs in cystic fibrosis, an autosomal recessive disease that can present with recurrent pneumonia and chronic diarrhea. However, neither *Pneumocystis pneumonia* nor lymphadenopathy is expected in patients with cystic fibrosis.

(Choice B) Low B cell concentrations are a feature of X-linked (Bruton) agammaglobulinemia (XLA). Infants with XLA have recurrent bacterial sinopulmonary infections, absent lymphoid tissue, and low serum immunoglobulin concentrations.

(Choice D) Defects in ciliary components such as dynein cause primary ciliary

lymphocyte deficiency. HIV infection presents with a normal absolute lymphocyte count and **CD4+ T lymphocyte deficiency** as most HIV replication occurs in CD4+ T cells. Because maternal antibodies may be present in HIV-negative children age <18 months, DNA PCR testing of blood can confirm the diagnosis. After age 18 months, persistence of HIV antibody is confirmatory of infection.

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(Choice B) Low B cell concentrations are a feature of X-linked (Bruton) agammaglobulinemia (XLA). Infants with XLA have recurrent bacterial sinopulmonary infections, absent lymphoid tissue, and low serum immunoglobulin concentrations.

(Choice D) Defects in ciliary components such as dynein cause primary ciliary dyskinesia. Affected patients have recurrent ear, nasal, and sinus infections and pneumonia due to bacterial infections. Failure to thrive, diarrhea, and *Pneumocystis* pneumonia are not seen in ciliary dyskinesia.

(Choice E) Adenosine deaminase deficiency is one of several gene defects resulting in severe combined immunodeficiency. Affected infants have profound lymphopenia, recurrent infections, and failure to thrive.

(Choice F) Impaired phagocytosis is seen in chronic granulomatous disease, which presents with recurrent, severe infections due to catalase-positive bacterial (eg, *Staphylococcus aureus*, *Serratia*) and fungal (eg, *Aspergillus*) organisms.

Educational objective:

HIV infection in infancy presents with failure to thrive, lymphadenopathy, and opportunistic infections (eg, *Pneumocystis* pneumonia, severe thrush). Selective loss of CD4+ cells is suggestive of HIV, and PCR reaction testing confirms the diagnosis.

References:

1. [Prevention of perinatal transmission of human immunodeficiency virus.](#)