

## Medical Quiz

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A four-week old Yemeni male infant presented after birth with symptoms and signs of duodenal obstruction, which required surgical intervention. The patient did well after surgery until 3 weeks postoperatively, when he developed difficulty of feeding, weight loss and loose motion. Blood test revealed lymphocytopenia. The patient had a chest X-ray, please answer the following questions.



- Q1.** Describe the X-ray findings.
- Q2.** What is the diagnosis?
- Q3.** Mention one associated condition.

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## **Answers to Medical Quiz**

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**A1.** Chest X-ray revealed absence of mediastinal thymus shadow.

**A2.** Absence of the thymus gland.

**A3.** Hypoparathyroidism.

### **DISCUSSION**

Immunodeficiency disorders in neonate are included in a group of conditions causing defects in immune cells function. Some disorders are common such as acquired immune deficiency syndrome (AIDS), while others are rare such as congenital absence of the thymus. These immunodeficiency disorders may affect single cell or more than one component of the immune system. Lymphocyte defects can be classified from functional point of view into three groups: humoral, cellular or combined immunodeficiencies<sup>1</sup>.

In most cases, the etiology of immune function defects in children could be due to viral bacterial or fungal infection and could be due to the side effects of drugs (secondary causes). Non infectious causes usually attributed to congenital absence of the thymus or genetic disorders (primary causes)<sup>1,2</sup>.

The signs and symptoms for immune defect due to absence of thymus include the following:

- T-cell deficiency and impaired antibody synthesis leading to recurrent bacterial, viral and fungal infection
- Failure to thrive
- Hepatosplenomegally
- Chronic diarrhea
- Other symptoms and signs include hypoparathyroidism, which depend on the association with other syndromes such as DiGeorge syndrome, which affects an infant's immune system and the function of parathyroid glands<sup>3,5</sup>.

Most cases of immune defects due to absence of thymus gland can be diagnosed on history of recurrent infections commonly affecting the lungs, clinical examination, blood tests, CT scan or MRI and chest X-ray<sup>6</sup>.

The management of these cases starts with a complete blood count, a manual differential of the cells. When lymphocytopenia (less than 2000/mm<sup>3</sup>) is persistent, the infant should be studied by flow cytometry and immunoassay. Recognizing lymphocytopenia can result in

early diagnosis of immunodeficiency disorders even at birth. The patient should be protected from infection by giving immune globulins prior to transplantation by bone marrow<sup>7,8</sup>.

The outcome of these cases depends on the timing of the diagnosis and the presence or absence of associated syndromes. Most deaths result from opportunistic infections commonly due to cytomegalovirus and adenovirus acquired before bone marrow transplant<sup>7,8</sup>.

## CONCLUSION

**Absent thymus is a rare cause of immunodeficiency disorders in neonates and children. Features of recurrent infections with lymphocytopenia are the usual presentations. The diagnosis can be confirmed after the blood test by CT scan or MRI and chest X-ray. Most cases require bone marrow transplant and they are at risk of death before transplant by opportunistic infections.**

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