Two Cases Wıth Tyrosine Kinase 2 Deficıency :

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About a quarter of the word's population is infected with Mycobacterium tuberculosis, but this bacterium causes tuberculosis in less than 10% of infected individuals. In the countries in wich tuberculosis is highly endemic, primary tuberculosis is particulary common in the children and adults. Clinical and epidemiological studies suggest that tuberculosis in humans has a strong genetic basis. Autosomal recessive (AR) complete interleukin-12 receptor β 1 (IL-12R β 1) and tyrosine kinase 2 (TYK2) deficiencies are the only two inborn errors of immunity reported to date to underlie primary tuberculosis in otherwise healthy patients in two or more kindreds (1,2). Inherited IL-12R β 1 and TYK2 deficiencies impair both IL-12– and IL-23–dependent IFN-g immunity and are rare also causes of tuberculosis (3,4).

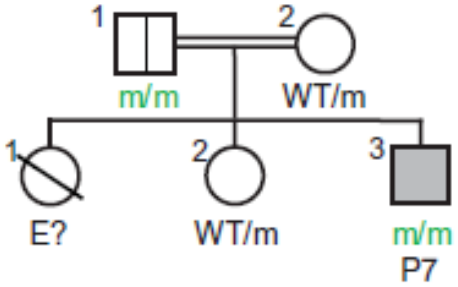
Case 1:

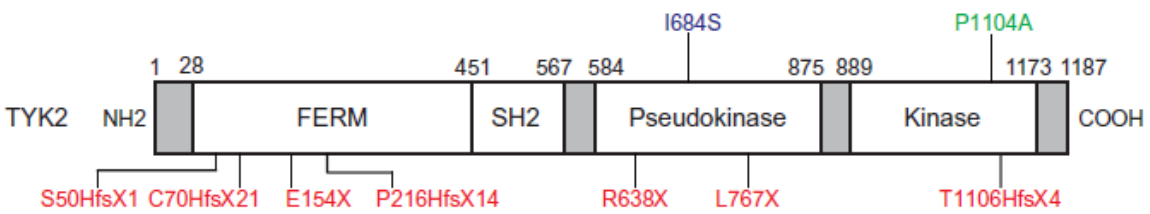
A 19 years old man from consanguineous marriages applied to chest disease. He is mainly complain cough for last 2 months. He has complaint of fever (39 °C). His weight loss for last 2 months. There is not any features of his medical history. But the patient's first admission to the hospital was 15 years old. This patient hospitalized with a diagnosis of pneumonia.

At physical examination: Respiratory system oscultation was normal. And also cardiovascular system was normal. There wasn't any abnormal finding. At Laboratory tests: Wbc: 8100 mm3 , Hb: 9,5 gr/dl , Crp: 63 mg/L , sedimantation rate 89 mm/H. Immunoglobulin levels are calculated as; IgE: 42,4 IU/ml, IgA: 334 mg/dl, IgM: 48 mg/dl, IgG: 1370 mg/dl. Lymph node biopsies were performed and obtained as a result of granulomatous inflammation and caseification necrosis. We started treatment with the diagnosis of miliary tuberculosis. Patient received; Isoniazid rifampicin, ethambutol, pyrazinamide treatment. After treatment clinical findings improved and weight gain normalized with antituberculosis treatment.

Genetic analysis:

HomozygousTYK2 mutation (P1104A) was detected in the genetic analysis;





Case 2:
A 46 years old, consanguineous marriage man. This patient is the father of our first patient's father. In his medical genetic research tyrosine kinase deficiency was detected. There is not any clinical findings: Espicially he has not got neither history of any infections nor tuberculosis. Immunological evaluation was normal except low IgM level like his son. Radiological exam was non-spesific. ARB staining and culture for TB were normal.

Results:
In cases with suspected tuberculosis, a case was presented to emphasize the importance of further investigation of tuberculosis susceptibility genes, even if there was no previous history of suspected immunodeficiency.

Figures:

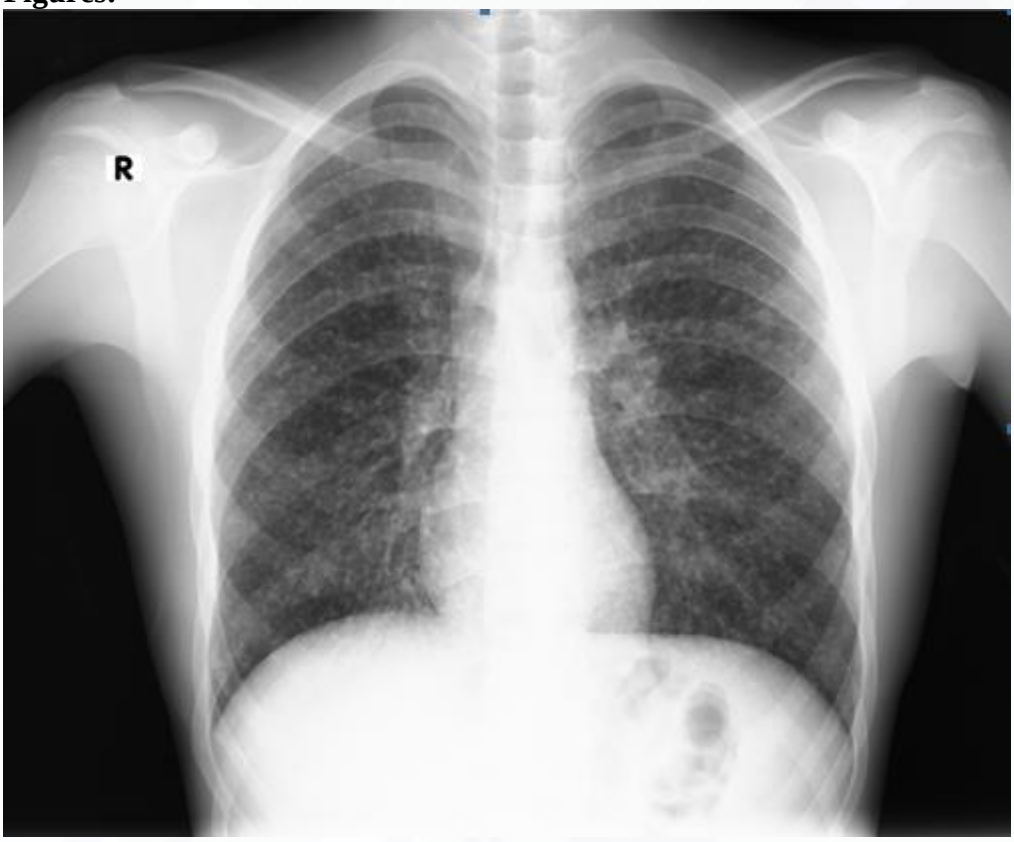


Figure 1: Posteroanterior chest graphy: Miliary lesions was seen.

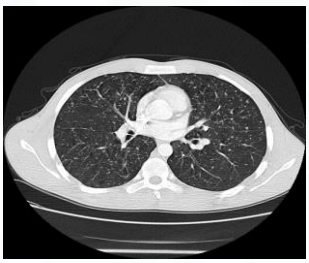


Figure 2: CT scan of lung: Miliary lesions was seen at axiel images.

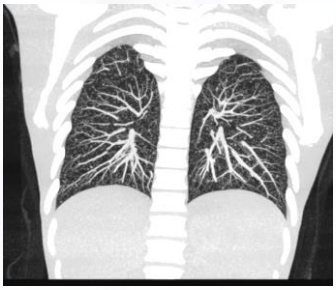


Figure 3: CT scan of lung : Miliary lesions was seen at coronal images.

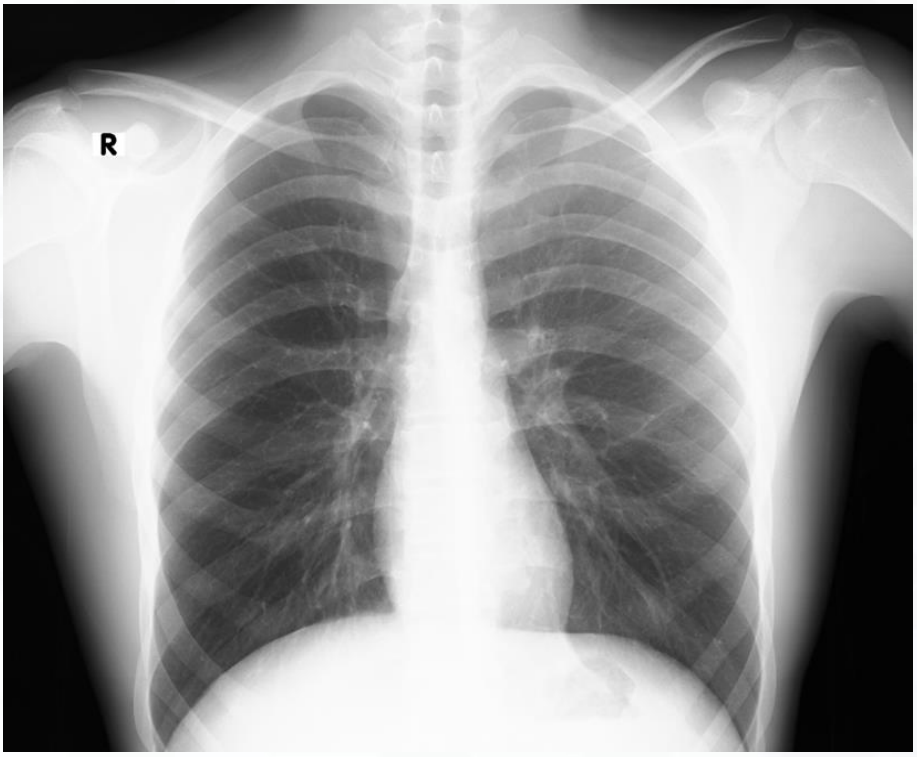


Figure 4: Posteroanterior chest graphy after treatment: Normally findings detected.

References:

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