Evaluation suggested

- Serum immunoglobulins and B cell counts.
- Humoral or antibody response to vaccines (post-vaccination antibody titres).

T Cell/Combined Defect

- Fungal and/or viral infections (e.g. *Aspergillus*, *Candida*, *Cytomegalovirus*) point towards an underlying cellular defect.
- Severe forms of T cell defects have an onset in early infancy (e.g. severe combined immune deficiency).
- Milder forms may have an onset later in life (e.g. combined immune defect due to LRBA deficiency, etc.).

Evaluation suggested

T cell counts (CD3 counts), serum immunoglobulins.

Phagocytic Defect

- Suppurative infections (complicated pneumonia—empyema/lung abscess, hepatic abscess, suppurative lymphadenitis) are seen in phagocytic defect.
- Onset—severe forms present in infancy, while milder forms may present later in life.

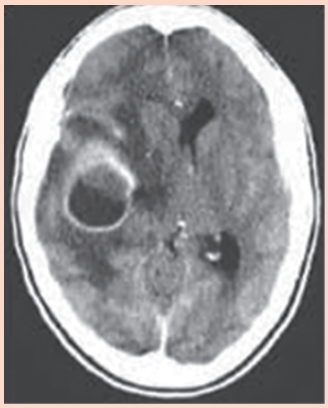
Evaluation suggested

Look at the absolute neutrophil counts. Neutropenia may point towards severe congenital neutropenia/cyclic neutropenia. High neutrophil counts must make one think of chronic granulomatous disease (*nitroblue tetrazolium test and dihydrorhodamine test are the screening tests for chronic granulomatous disease*).

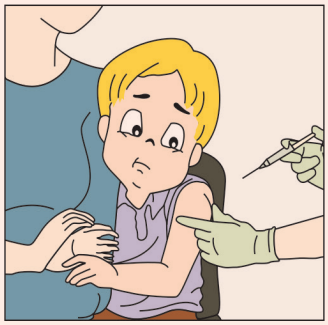
Complement Deficiency

- Recurrent infections with encapsulated bacteria (*S. pneumoniae*, *H. influenzae*) can be noted in complement deficiency.
- Early onset of autoimmune diseases can be a feature of complement deficiency (e.g. onset of lupus before the age of 5 years is seen in complement C1q deficiency).

Failure to thrive—not gaining weight and height as per the age norms



Repeated abscess formation (liver abscess, brain abscess)



Infections warranting multiple hospitalizations

Database
3-year-old boy with recurrent pneumonia
↓
Absent tonsils and lymph nodes
↓
Low immunoglobulins and absent B cells
↓
Diagnosis: X-linked agammaglobulinemia

Message: Recurrent pneumonia—serum immunoglobulins must be tested.

A Quick Look into the Disease

X-linked Agammaglobulinemia (Previously called Brutons Agammaglobulinemia)

- Boys are affected.
- Recurrent pneumonia, otitis media, skin infections, diarrhea.
- Absent tonsils.
- Low immunoglobulins and absent B cells.
- Mutation in the *BTK* gene.

Case 2

28-year-old gentleman was referred by the pulmonologist for immunological evaluation. He had had repeated episodes of pneumonia requiring 7–10 days of antimicrobials during each episode. He had 2–3 such episodes every year for the past 8 years.

Past History

Repeated episodes of ear discharge from the age of 10. Recurrent sinusitis from the age of 15–3 to four episodes/year (was said to have allergic rhinitis!).

He had been extensively evaluated before being referred.

CT chest done thrice revealed consolidation involving different lobes during these episodes of pneumonia. Latest CT chest showed changes of bronchiectasis in the lower lobes. He had undergone bronchoscopies but no

3. A 6-year-old girl presented with fever and tender hepatomegaly. USG abdomen showed multiple *liver abscesses*. Pus was aspirated and culture grew *pseudomonas*.
4. A 7-year-old child presented with third episode of pneumonia. Chest radiograph showed pleural effusion. Pleural aspirate revealed frank pus. Diagnosed to have *empyema* and inter-coastal drainage (ICD) tube placed.

Would you investigate such cases for immune deficiency in day-to-day practice?

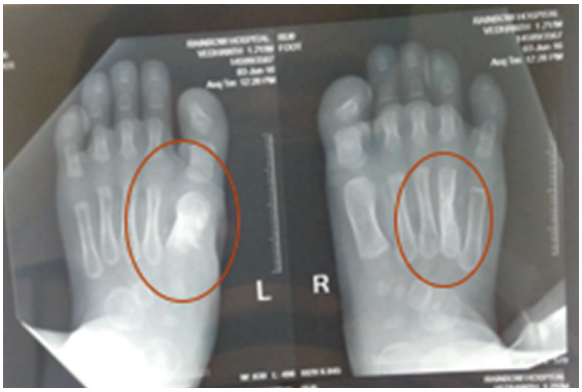


Fig. 7.1: Radiographs of the feet showing osteomyelitis affecting multiple metatarsal bones

Investigations in these cases				
	Case 1	***Case 2***	***Case 3***	***Case 4***
Hb (g/dl)	9	8.5	10	9.5
TC (per mm^3)	20,000	18,000	15500	17300
DC	$N_{75}L_{15}$	$N_{70}L_{18}$	$N_{80}L_{16}$	$N_{78}L_{14}$
PC (per mm^3)	600,000	650,000	800,000	550,000
IgG (mg/dl)	2100	1750	3200	1900
IgA (mg/dl)	150	220	345	180
IgM (mg/dl)	205	154	235	160