

History and Clinical Examination

Nomhle Shongwe arrives to see Dr Goodheart for a scheduled follow-up appointment with her 3-year old son Anathi. He presented the previous week, at the paediatric clinic, with a history of recurrent infections. Dr Goodheart begins the consultation by taking a detailed history.

General history:

Nomhle was healthy during her pregnancy with Anathi and tested **negative for HIV**.

The birth history was normal. He was born at **term, weighing 3.4kg**.

He was **breastfed** for the first year of life.

He received **standard immunisations**, and **no side effects** were observed **to live vaccines**.

Dr Goodheart excludes a history of **dissemination after BCG vaccination** and **infection with environmental species of Tuberculosis**, both of which are relevant concerns in South Africa.

Social history:

The Shongwe family lives in a two-bedroomed **house with running water and electricity**.

Both Anathi's parents are **employed** and neither of them **smoke** or **drink alcohol**.

During the day Anathi is **cared for at home**, by his grandmother, with his one-year old healthy sister Thabisa.

Anathi has **not travelled** outside his home town in his three years of life.

The family has a **small dog** and no cats.



Authors

Dr Walter Liebrich, PhD

Dr Shaunagh Emanuel (MBChB)

Dr Monika Esser, MMed Paed (Rheum)

Reference:

10 Warning Signs Children and Adults.
Jeffrey Modell Foundation Medical Advisory Board. Ten warning signs of primary immunodeficiency. <http://www.info4pi.org> (accessed 23 September 2014).

Family History:

Anathi's **parents are not closely related** and there is no family history of **autoimmunity** or **cystic fibrosis**.

No family members have tested positive for **HIV**.

No close family members have been diagnosed with **tuberculosis** or **recurrent meningococcal infections**.

Dr Goodheart enquires about other family members with a **similar disease pattern**, in particular **death of children in infancy**.

Anathi's uncle on his mother's side died from recurrent infections as a young boy.

Infection History:

Dr Goodheart looks out for **SPUR** warning signs (**severe, persistent, unusual, or recurrent infections**).

Anathi did not have any of the following: recurrent **ear infections**, recurrent **deep skin or organ abscesses**, **persistent oral thrush**, **fungal skin infections** or **septicemia**.

He has not suffered from **meningitis** or **encephalitis**.

Anathi has, however, had two serious **sinus infections**.

He also developed **pneumonia on two separate occasions**. According to the referring doctor, oral antibiotics had little effect and **he required intravenous antibiotics** to clear these. He has also suffered from **intermittent diarrhoea**.

Early onset of severe infections is more commonly associated with serious primary immunodeficiencies (PIDs). In Anathi's case, he began to suffer from severe recurrent

infections **soon after he was weaned from breast milk**.



'Signature organisms'

PIDs, depending on the **predominant immune defect**, present with susceptibility to infections caused by **'signature' organisms**:

T cells – *Pneumocystis jirovecii* (previously *P. carinii*), mycobacteriae, *Cryptococcus neoformans*, herpes viruses;

B cells (antibody related) – *Streptococcus pneumoniae*, *Haemophilus influenzae*, *Giardia lamblia*, enteroviruses, staphylococci, *Campylobacter* spp;

Complement – *Neisseria* spp (Meningococcus) – important in South Africa;

Phagocytes – staphylococci, atypical mycobacterial species, *Aspergillus* spp, *Burkholderia cepacia*,

Sputum cultures were positive for *Streptococcus pneumoniae* on both occasions when Anathi was previously diagnosed with pneumonia.

This is suggestive of a possible antibody problem.

Dr Goodheart considers that Anathi may have a PID in view of his infection history and family history.

Clinical Examination:

When Dr Goodheart **examines** Anathi she pays careful attention to the following:



Anathi is **failing to thrive**. She knows this is not always evident. Despite being small and underweight, he has normal developmental milestones. He looks **chronically ill** and **pale**. Early **clubbing of his nails** is present, indicating possible chronic lung disease. He has no obvious ectodermal defects - his **hair** is not excessively dry and his **teeth** look normal. He has no suggestive **skin** features such as severe eczema, abscesses, petechiae, telangiectasia or mucosal ulcerations or thrush. He is **not dysmorphic** and has no abnormal features of obvious syndromes (e.g. Di George- 22q deletion). On examination of the throat, **lymphoid tissue is notably absent and he has no tonsils** although he has suffered from recurrent sinusitis and he has not had a tonsillectomy. Surprisingly his **eardrums** are not perforated. (Subclinical otitis media is therefore unlikely). He has normal **heart** sounds and his blood pressure is within normal limits. There are no abnormal findings on neurological examination and he is **not ataxic**. His axillary **temperature is elevated to 38.5°C**. His **respiratory rate is increased** and he coughs intermittently. On chest examination **bilateral crepitations** are present.

Dr Goodheart makes the diagnosis of pneumonia and arranges for immediate admission. She sends off preliminary blood tests to the laboratory. On the way to the ward Anathi visits the radiology department for a chest X-ray.

[In the third article of this series we will continue to follow Anathi's progress and learn more about investigations in PID.](#)