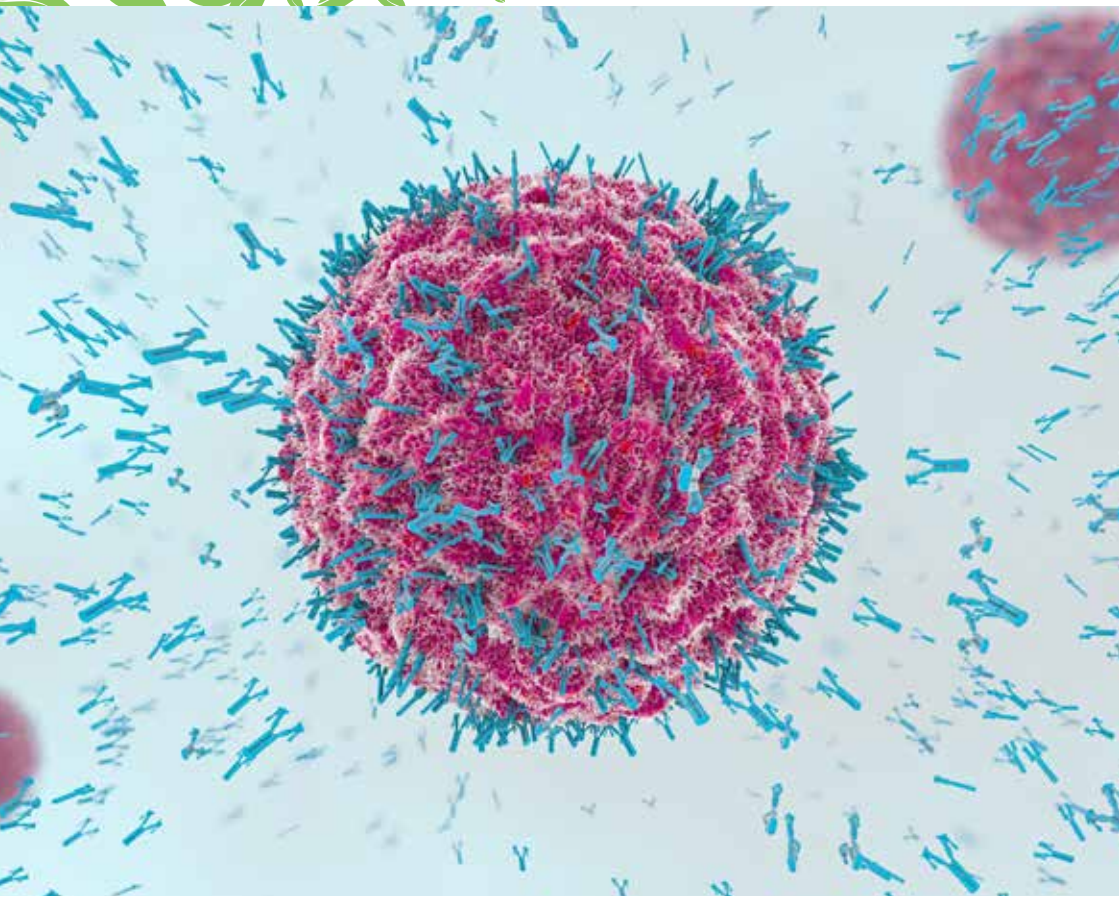
Diagnosing Patients with a Primary Antibody Deficiency



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Purpose

The purpose of this document is to give GPs the understanding they need to enable the identification of patients who may have an undiagnosed Primary Antibody Deficiency (PAD), to help determine what initial tests might be required, and to understand when a patient should be referred to a Clinical Immunologist.

What is a Primary Immune Deficiency?

The term Primary Immune Deficiency (PID) means a disease that has happened because of a fault (usually a genetic fault) in the body's ability to either produce or maintain a complete immune system. Just as there are many different parts of the immune system, there are also many different types of Primary Immune Deficiency.

Primary Antibody Deficiency (PAD) means that the immune system cannot make antibodies. The inability of the immune system to make antibodies is not the same in every person who has an antibody deficiency.

Prevalence

The prevalence of PAD is said to be around **1:10,000**. An average GP practice could expect to have at least one patient with a Primary Immune Deficiency within their patient list. Although PAD is rare, diagnosis makes a big difference to the patient in terms of preventing organ damage, especially bronchiectasis.

Would you consider any of your patients as a possible PAD?

Most PAD patients are diagnosed in their twenties and thirties. Many people believe that patients with an undiagnosed PAD will succumb to the condition very soon after the onset of disease, but for the majority, this is not the case at all. In fact PADs usually present as "revolving door" patients, with chronic infections of the upper respiratory tract that will not resolve despite their GP's best efforts.

Diagnosing Patients with a Primary Antibody Deficiency

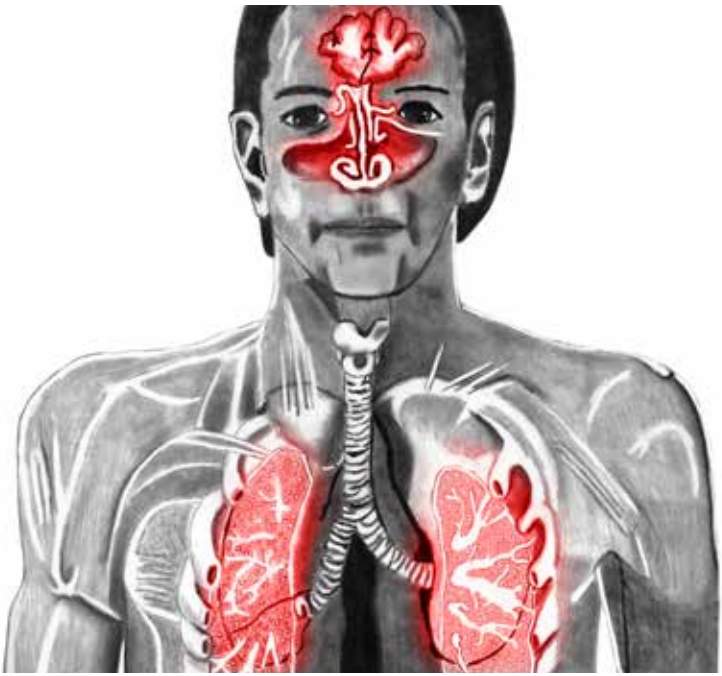
Would you consider identifying any of your patients who have had any of the following?

(These are the typical signs that a PAD should be considered)

- Patients needing more than three prescriptions for antibiotics in the last year.
- Patients with chronic infections such as sinusitis, tonsillitis, otitis media, or recurrent boils.
- Infections that resolve with antibiotic therapy but return soon after the course is completed.
- Requirement for frequent courses of oral antibiotics.
- Needed intravenous antibiotics.
- Have had pneumonia.
- Children with failure to thrive.
- Patients diagnosed wit bronchiectasis.
- Patients who have had particularly severe, unusual and persistent infections.
- Patients who have enlargement of liver, spleen, and/or other lymphoid tissue abnormalities.

How do PADs present?

Most patients with an undiagnosed PAD will present with recurrent infections. The most common presentation is of upper and lower respiratory infections which sometimes resolve with treatment but keep returning, despite appropriate antibiotic therapy. Patients with a Primary Antibody Deficiency often do not “look” unwell and this can be confusing. Many patients living with a PAD have a low normal temperature.



Are PADs difficult to diagnose?

PADs are sometimes difficult to assess since much of the presentation is with “normal” infections. However, these diseases are relatively easy to diagnose and patients diagnosed early after the onset of disease will often be able to lead near to normal lives.

What tests should a GP order?

GPs who suspect that a patient may have a PAD should order the following:

1. Full blood count. Many PADs will be associated with low cell numbers which are often autoimmune, including neutrophils, platelets etc.
2. IgG, IgA, IgM.
3. Lack of antibodies to childhood infections such as chicken pox.
4. Low or absent antibodies to vaccinations (childhood or otherwise e.g. rubella, hepatitis B)
5. Microbiology. This is a simple test which could provide much information to guide further investigations and management. This may be in the form of sputum, cough swabs, throat swabs, etc.

When to refer to a Clinical Immunologist?

If any of the tests are abnormal, the patient must be referred to a Consultant Clinical Immunologist without delay. However, even if these tests come back within normal limits and the patient continues to present with symptoms that imply PAD, they should be referred. This is because more subtle Primary Antibody Deficiencies which require more sophisticated testing can also cause serious illness but cannot be identified by tests available to GPs.

If unsure about the above investigations, please discuss your patient with a consultant immunologist, even prior to undertaking any investigations.

How to find a Clinical Immunologist?

HMC and Sidra Medicine. Patients have to be referred out of the immediate area in order to obtain appropriate diagnosis and treatment.

NOTES:

1. Although all care has been taken, this booklet is a general guide only which is not intended to be a substitute for individual medical advice/ treatment. Allergy and Immunology Awareness Program in Qatar and UK-Primary Immunodeficiency Patient Support (UK-PIPS) expressly disclaim all responsibility (including negligence) for any loss, damage or personal injury resulting from reliance on the information contained.

2. We would like to thank UK-Primary Immunodeficiency Patient Support (UK-PIPS) for giving us permission to include their patient education booklet in our product.

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