

References:

For references please contact UKPIPS

More Information

For further information about this leaflet or any PID issue, contact UKPIPS at:

T: 01386 421228. Mob: 07531 076409

W: www.ukpips.org.uk

E: info@ukpips.org.uk.

UKPIPS: De Montfort House 7E Enterprise Way
Vale Business Park Evesham



UKPIPS
De Montfort House
7e Enterprise Way
Vale Business Park
Evesham
Worcestershire
WR11 1GU

Tel: 01386 421228 & 07531 076409

Email: info@ukpips.org.uk

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For details of information sources,
contact info@ukpips.org.uk



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DIAGNOSING PATIENTS WITH A PRIMARY ANTIBODY DEFICIENCY



UK
Primary
Immune-deficiency
Patient Support

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Purpose

The purpose of this document is to give GP's the understanding they need to enable them to identify patients who may have an undiagnosed Primary Antibody Deficiency (PAD) and then to know what initial tests they can undertake and when to refer their patient to a Clinical Immunologist.

Prevalence

The prevalence of PAD is said to be 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 die very soon after the onset of disease, but for the majority, this is not the case at all. In fact PAD's usually present as "revolving door" patients, with **chronic infections of the upper respiratory tract** that will not resolve, despite their GP's best efforts.

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, chest infections 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 with 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 infection which sometimes resolve with treatment - but keeps returning, despite appropriate antibiotic therapy. Patients with a Primary Antibody Deficiency often do not "look" unwell and this can be confusing. They do not look unwell because their immune system is not functioning appropriately. Anecdotally, UKPIPS understands that many patients living with a PAD have a low normal temperature, many therefore being febrile at 37.0°C

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:
- Full blood count. Many PADs will be associated with low cell numbers which are often autoimmune including neutrophils, platelets etc.
- IgG, IgA, IgM.

- Lack of antibodies to childhood infections such as Chicken Pox.
- Low or absent antibodies to vaccinations (childhood or otherwise - e.g. Rubella, Hepatitis B)
- 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 you are unsure about the above investigations then please discuss your patient with a Consultant Immunologist, even prior to undertaking any investigations.

How to find a Clinical Immunologist

Consultant Clinical Immunologists work in specialist regional centres and patients have to be referred out of the immediate area in order to obtain appropriate diagnosis and treatment. A list of these specialist centres can be found on the UKPIPS, BSI and UKPIN Websites.

Funding concerns should never prevent a GP from either testing for a PAD or referring a patient for testing since, once diagnosed, these patients come under NHS England Specialist Commissioning arrangements.