NIHR | BioResource

PID – Primary Immunodeficiency

NIHR BioResource – Rare Diseases study project

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Summary

Primary Immunodeficiency (PID) is a large group of genetic disorders that affect more than 1% of the UK population and can underlie susceptibility to debilitating infection, auto-immune disease and some cancers. It can have devastating impact on the lives of patients and their families. However, PID is often unrecognised because of variations in the severity and types of symptoms that can delay diagnosis and the implementation of appropriate care.

Prof. Ken Smith, PID project Lead

We recently completed a pilot study that combined the genetic and clinical data of PID patients to identify new genetic causes in order to improve diagnosis for affected individuals. We are extending this work to integrate genetic, clinical, and laboratory measures of immune function from PID patients, and use this information to unravel the biological mechanisms that control our immune cells and can result in failure of our immune systems to operate normally. Ultimately this will inform diagnosis and enable the appropriate use of therapeutics to transform the lives of this significant patient population. Our findings may also have impact for other areas of medicine such as autoimmunity and cancer, where immune cells play a part in the disease process.

Recruitment Criteria

Inclusion

- Suspected primary immunodeficiency AND / OR
- Recurrent / protracted or unusual infection

A patient's relative(s) (affected, as above; or unaffected) can be recruited to the PID project.

Exclusion

- Secondary immunodeficiency resulting from previous immunosuppressive / chemotherapy treatment preceding the onset of infections
- Secondary immunodeficiency resulting from renal / gut / other loss of immunoglobulin and/or lymphocytes