# Activated PI3 Kinase Delta Syndrome

## Contact details
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## Samples required
- 5ml venous blood in plastic EDTA bottles (>1ml from neonates)  
- Prenatal testing must be arranged in advance, through a Clinical Genetics department if possible.  
- Amniotic fluid or CV samples should be sent to Cytogenetics for dissecting and culturing, with instructions to forward the sample to the Regional Molecular Genetics laboratory for analysis  
- A completed DNA request card should accompany all samples

## Patient details
To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address and ethnic origin), details of any relevant family history and full contact details for the referring clinician

## Introduction
Activated PI3 Kinase Delta Syndrome (APDS) is a newly discovered cause of Primary Immunodeficiency (PID). It is an autosomal dominant disorder due to gain of function in the catalytic subunit phosphoinositide 3-kinase δ (p110δ) encoded by the PIK3CD gene.  

Activation of p110δ can induce cell growth, proliferation and many other cellular functions. APDS is a clinically heterogeneous condition with variable penetrance among affected individuals. The most common manifestation is susceptibility to recurrent infections, progressive lung disease, splenomegaly and lymphoproliferative manifestation with mainly antibody deficiency.

Three recurrent heterozygous gain-of-function mutations in exon 8, 13 and 24 of the PIK3CD gene have been reported in patients affected by APDS; c.1002C>A p.(Asn334Lys); c.1573G>A p.(Glu525Lys) and c.3061G>A p.(Glu1021Lys) respectively.

## Referrals
Confirmation of diagnosis in a patient with Activated PI3K-delta syndrome (APDS) / Primary B cell immunodeficiency.  
Family members with previously identified PIK3CD mutation

## Prenatal testing
Prenatal diagnosis may not be appropriate - please contact the laboratory to discuss families in whom specific mutations have been identified or in whom appropriate family studies have been undertaken.

## Service offered
Testing is performed by direct Sanger sequence analysis of the coding exons 8, 13, 24 of the PIK3CD gene.

## Technical
Mutation screening is carried out by direct sequencing analysis.

## Target reporting time
The target reporting time is 4 weeks. Please contact the laboratory for urgent cases.