

# POD. Phenotyping Overgrowth Disorders

NIHR BioResource – Rare Diseases study project

**Lead Investigator: Dr Alison Foster**

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## Summary

Childhood overgrowth disorders are a group of rare genetic conditions associated with increased growth, learning disability, congenital abnormalities, and a wide range of other medical problems. Some children have an increased risk of developing tumours.



**Dr Alison Foster, POD project lead**

In this project, Dr Foster will establish a group of patients who agree to donate samples that can be used for future research on overgrowth disorders. She will also identify groups of patients for further genetic investigations. This work could lead to new ways of managing these conditions and a group of patients suitable for trials of new drugs.

Patients joining the study will undergo a full clinical examination and their medical notes will be reviewed. The results of previous tests and scans will be included in the research. Participants will be asked for a blood or saliva sample. They may also be asked to provide a skin sample, for example if there is overgrowth of a single region of the body.

Dr Foster is based at the Birmingham Women's and Children's NHS Foundation Trust. The RD-TRC Fellowship was awarded in 2014. The full title of her study is 'Phenotyping of rare genetic overgrowth disorders (POD) study'.

## Recruitment Criteria

Recruitment of trio's (proband plus parents)

### Inclusion

Height or OFC > 2SD above the mean (~>98th centile)

**AND** Dysmorphic facial features, developmental delay, congenital abnormality or childhood tumour

OR Height or OFC >3SD above the mean (~>99.6th centile)

OR Segmental overgrowth or hemihypertrophy

OR A likely pathogenic variant in a known overgrowth gene

### Exclusion

Tall stature or macrocephaly due to other cause not considered to be a primary overgrowth disorder (e.g. Marfan syndrome, acromegaly).