NIHR | BioResource

# ACH. Achondroplasia and Hypochondroplasia

NIHR BioResource – Rare Diseases study project

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## Summary

Achondroplasia and Hypochondroplasia combined are the commonest of the skeletal dysplasias- a group of more than 400 conditions causing changes in bone growth resulting in short stature. More than 1 in 20,000 people have one of these diagnoses. Both conditions are due to changes in the FGFR3 due which cause shortening of the long bones of the arms and legs and alterations in the growth of the bones of the skull and the spine. People with achondroplasia and hypochondroplasia are at risk of a number of neurological, orthopaedic and respiratory complications including cervicomedullary compression, long bone deformity and sleep disturbed breathing. They frequently require surgical intervention to treat these. In general people with hypochondroplasia have fewer complications than those with achondroplasia.

Despite the fact that most people with achondroplasia have one of a very small number of changes in the FGFR3 gene there is enormous variability in the pattern of problems that people with these conditions have. There is poor understanding of why this should be the case.

The purpose of recruiting people to this cohort in the Bioresource - Rare Diseases Study is to facilitate further studies to improve our understanding of the variability we see in people with achondroplasia and to support the development of new therapies.

## Recruitment Criteria

### Inclusion

Patients with:

- A clinical and radiographic diagnosis of achondroplasia or hypochondroplasia

- A variant in the FGFR3 gene known to be associated with either achondroplasia or hypochondroplasia

## Exclusion

Patients with an alternative skeletal dysplasia diagnosis.