NIHR | BioResource

AMC – Arthrogryposis

NIHR BioResource – Rare Diseases study project

Lead Investigators:
Dr Andrea Jester & Dr Julie Vogt

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Summary

Arthrogyposis (multiple curved joints) is a term used to describe group of disorders in which individuals have congenital joint contractures. The incidence is between 1 in 5000 and in 3000 live births, with approximately 200 babies born in the UK each year. Babies with this condition warrant a full clinical assessment soon after birth so that early specialist intervention with physiotherapy and surgery can be considered with the aim of achieving optimal joint function. In addition, children may benefit from orthotics, occupational therapy, speech and language assistance, further surgical evaluation and follow up and support at school and within the home.

Dr Andrea Jester (top) & Dr Julie Vogt, AMC project Leads

Extensive phenotypic and genetic heterogeneity hampers the ability to determine an underlying clinical and molecular diagnosis, which may assist in patient management and enable accurate genetic counselling for families, and patients often undergo multiple costly and invasive investigations. It is clear that the varied presentation of this condition and its rarity leads to delay with establishing an early diagnosis and in the provision of an individualised management plan.

Birmingham Women's and Children's Hospital Arthrogryposis team is leading an NIHR (National Institute for Health Research) BioResource Arthrogryposis cohort with colleagues from Newcastle, Manchester and Great Ormond Street. Firstly, we have designed a purpose built database to facilitate the collection of detailed phenotypic information to broaden our understanding of the presentation and associated complications in our arthrogryposis cohort and improve individualised patient care. We plan to develop and validate treatments in these patient subgroups, thus improving the care for patients with arthrogryposis and their families. Secondly DNA will be stored from participants. Single nucleotide polymorphism analysis will be undertaken on probands, their parents and other relatives recruited to the study. In the future we would like to improve rates of molecular diagnosis, identify new candidate genes

and study the effect of mutational burden in the arthrogryposis by analysing genomic data in conjunction with the phenotypic profile.

Recruitment Criteria

Inclusion

All patients born with joint contractures; 2 or more joint groups affected (syndromic and non-syndromic, uni- and bilateral, known or unknown cause or genetic diagnosis, post-mortem or pregnancy samples).

In addition to proband with arthrogryposis (multiple joint contractures), parents (affected or unaffected) and other affected relatives can be recruited to this AMC project.

Exclusion

Joint contractures that are not congenital (present at birth).
Less than 2 joint groups affected.