Experience from an interdisciplinary arthrogryposis clinic in the North of England

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Introduction:
Arthrogryposis is a descriptive term referring to multiple congenital contractures. More than 400 different conditions can present with contractures at birth. The underlying cause has now been identified in more than 150 of these conditions, but is still unknown for a large proportion of patients presenting with arthrogryposis. Because of the rarity and complexity of patients with arthrogryposis, coordinated care and management is often missing.

Methods:
We report about our 9 years experience of an interdisciplinary arthrogryposis clinic, set up to cover all aspects of diagnoses and care. The clinic provides: diagnostic work-up, treatment of associated anomalies and overall care co-ordination of paediatric patients with arthrogryposis.

Representatives from a patient association (TAG) can provide families with advice and support. Patients can access the service through referrals from a variety of specialties, eg. foetal medicine, paediatrics and paediatric neurology, orthopaedics and genetics.

Since its foundation the clinic witnessed the evolution of genetic diagnostic technologies, from single gene analysis, to gene panels for specific groups of diseases and exome or genome sequencing for more complex cases (Table 1). New sequencing technologies can contribute to an accurate and expert clinical evaluation and can increase the chance to provide a precise genetic diagnosis and a correct genetic counselling.

Results:
Table 2: Our patient cohort is summarized in this table. Nearly 60% have a recognized diagnosis: half of them (29%) have amyloplasia, in accordance with reported data (Hall J et al. 1984). The other diagnoses represent a wide range of different conditions, some represented in figure 2, 3 and 4.

Discussion:
An interdisciplinary approach helps to reduce the number of appointments, provide patients with arthrogryposis with more specific diagnoses, coordinated orthotic and surgical management and informed physiotherapy.