Handicap in Adults with Arthrogryposis is Severe, multifaceted, Partly Invisible and Varies by Genotype

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Introduction

Arthrogryposis multiplex congenita (AMC) is a disease characterized by the presence of at least two joint contractures at birth in different body areas. This study is the first to describe disability patterns of a cohort of adults with AMC.

Methods

Retrospective analysis of data for unselected persons with AMC referred to the French center for adults with AMC from 2010 to 2016. All underwent a pluri-professional systematic and comprehensive investigation of deficits, activity limitation, and participation restriction according to the International Classification of Functioning, Disability and Health and genetic analysis when indicated. Participants were divided by Amyoplasia and other AMC types.

Results

Muscle weakness

Joint stiffness

Respiratory impairment

Severe respiratory disorders were infrequent and were linked to *PIEZO2* mutations. Respiratory function was better with Amyoplasia than other AMC types (FVC 3.8 [3-4.3] vs 2.2 [1.4-2.7], $p<0.001$).

Swallowing and speech disorders

Prevalence	Total n (%)	Amyoplasia n (%)	Other types n (%)	p
Patients with a limited amplitude in mouth opening (< 3 fingers = 40 mm)	10 (24.4%)	6 (23.1%)	4 (26.7%)	ns
Patients with chewing problems	15 (36.6%)	8 (30.8%)	7 (46.7%)	ns
Patients with swallowing problems	5 (12.2%)	1 (3.8%)	4 (26.7%)	$p=0.034$
Patients with voice problems	5 (12.2%)	0 (0%)	5 (33.3%)	$p=0.002$
Patients with speech problems	4 (10.0%)	3 (11.5%)	1 (6.7%)	ns

Pain and psychological suffering

About 91% of patients complained of pain, other principle psychological problems were: anxiety (43%), fatigue (34%), difficulty in sexual life (24%), altered self-esteem (17%), and feeling of solitude (15%).

Activity limitations

As shown in fig, most patients had modified independence with FIM 113 ± 13.9 . Patients with Amyoplasia had lower FIM scores than patients with other types of AMC (110.4 ± 16.3 vs 118.1 ± 4.8 , $p=0.26$). Nearly all patients had high scores at the Cognitive FIM (34.8 ± 0.6).

Mobility

Most participants with AMC (88%) could walk independently. Gait disorders were not due to respiratory impairment but to skeletal problems and were always associated with Amyoplasia when severe.

Participation restriction

	Amyoplasia (n=28)	Other types (n=15)	p
Occupation			
Work	14 (50%)	8 (53%)	ns
Student	7 (25%)	3 (20%)	ns
unemployed	7 (25%)	4 (27%)	ns
Technical assistance	17 (61%)	4 (27%)	ns
Human help	12 (43%)	5 (33%)	ns
Driving	15 (54%)	13 (87%)	$p=0.03$

Conclusion

This study describes disability patterns of a cohort of adults with AMC by genotype. The handicap of adults with AMC is influenced by genotype, with an important invisible handicap.

Figure 1