Use of Aids, Assistive Devices and Adaptations by Individuals with Fibrodysplasia Ossificans Progressiva (FOP): 36-Month Results from a Global Natural History Study

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Mona Al Mukaddam,[1] Geneviève Baujat,[2] Aude Houchard,[3] Edward C. Hsiao,[4] Richard Keen,[5] Rose Marino,[6] Robert J. Pignolo,[7] Frederick S. Kaplan[1]

[1]Departments of Orthopaedic Surgery and Medicine, The Center for Research in FOP and Related Disorders, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA; [2]Département de Génétique, Institut IMAGINE and Hôpital Universitaire Necker-Enfants Malades, Paris, France; [3]Ipsen, Boulogne-Billancourt, France; [4]Division of Endocrinology and Metabolism, the UCSF Metabolic Bone Clinic, the Institute of Human Genetics, and the UCSF Program in Craniofacial Biology, Department of Medicine, University of California San Francisco, San Francisco, CA, USA; [5]Centre for Metabolic Bone Disease, Royal National Orthopaedic Hospital, Stanmore, UK; [6]Ipsen, Newton, MA, USA; [7]Department of Medicine, Mayo Clinic, Rochester, MN, USA.

Presenting author: Rose Marino (rose.marino@ipsen.com)

Background

- Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare genetic disorder.
- 97% of individuals with FOP have classic FOP, which is associated with an R206H mutation in activin A receptor type I (ACVR1; also known as activin receptor-like kinase-2).[1]
- FOP is characterised by episodic progressive heterotopic ossification (HO), often preceded by flare-ups.[2–4]
- HO develops into ribbons, sheets, and plates of extra bone throughout the body, progressively restricting movement.[4]
- Disability in FOP is cumulative, with most patients requiring the use of a wheelchair by their third decade of life and lifelong assistance in performing activities of daily living.[2,3]

Objective

To characterise the use of aids, assistive devices, and adaptations (AADAs) in individuals with FOP enrolled in a 36-month, global natural history study (NHS).

Methods

- A 36-month, prospective, protocol-specified, longitudinal NHS was conducted (NCT02322255).
- Individuals with FOP aged ≤65 years with a documented *ACVR1*^{R206H} mutation were eligible to participate.
- This analysis reports use of AADAs at Baseline and Month 36, and new-onset use at Month 36 of the NHS.
- The International FOP Association has compiled a list of AADAs that patients with FOP may use.
  - AADAs were grouped into 12 categories, and participants were asked to select AADAs used for daily living at NHS Day 1, and during clinic visits and/or telephone appointments at Months 6, 12, 18, 24, 30, 36.
  - New-onset AADAs were defined as AADAs used at the specified visit that were not used or not applicable at Baseline.
- Data are presented by AADA category.

Results

- A total of 117 participants were enrolled in the NHS, of whom 114 had an *ACVR1*^{R206H} mutation and provided Baseline data **(Table 1)**.
- Data on AADA usage at Baseline and Month 36 were available for 108 and 41 participants, respectively.
  - 37 participants provided data for new-onset AADA use at Month 36; the low patient number observed reflects the high rate of study discontinuation (71.1%), largely due to participants enrolling in interventional studies (53.5%).
- The number of AADAs used per participant per category at Baseline (mean [SD]) was lowest for sport and recreation adaptations (1.5 [0.6]) followed by care attendants (1.6 [0.7]), and highest for home adaptations (3.2 [2.6]) followed by bathroom aids and devices (2.9 [1.7]).
- Personal care tools/aids (71.3%) and bedroom aids and devices (73.2%) were the categories of AADAs used by the highest proportion of participants at Baseline **(Figure 1A)** and Month 36 **(Figure 1B)**, respectively.
- The 3 most common new-onset AADA categories between Baseline and Month 36 overall were care attendants, personal care tools/aids, and bathroom aids/devices (reported by >45% of participants) **(Figure 1C)**.

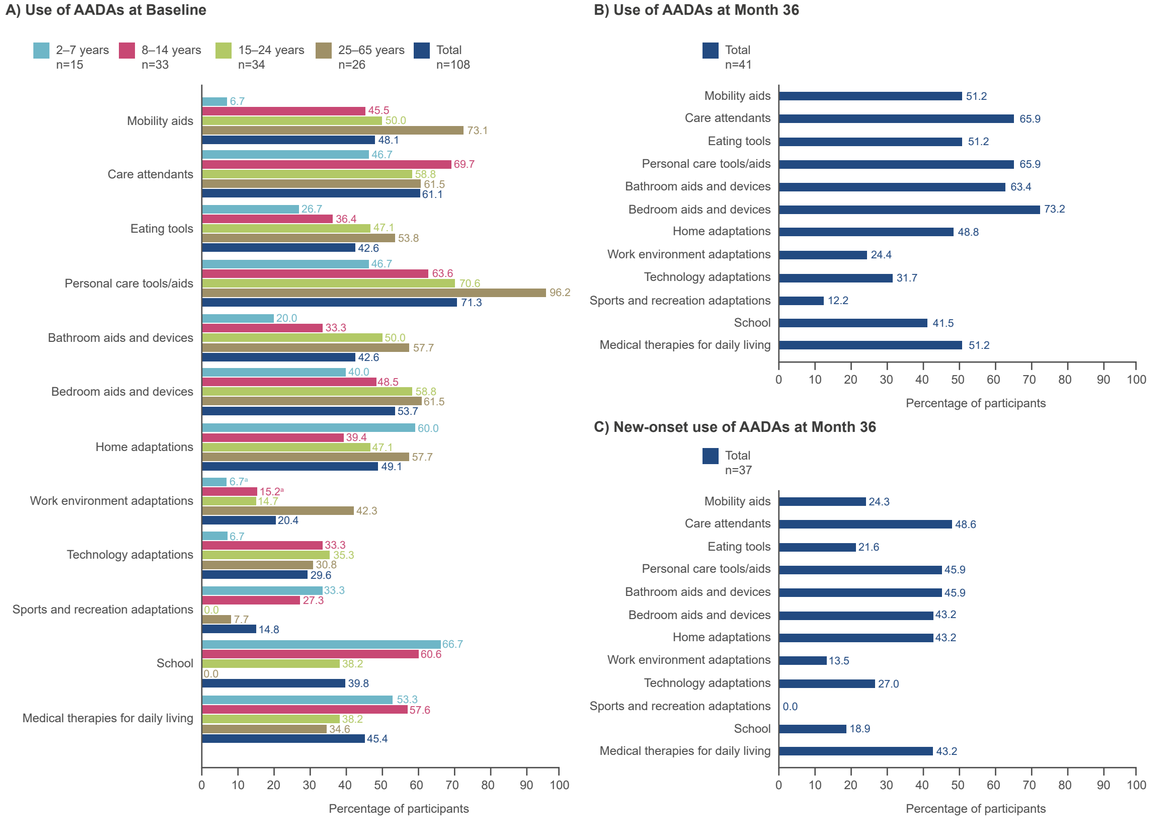
Table 1. Baseline demographics and characteristics

Characteristic	Patients who provided use of AADAs at Baseline					Patients who provided use of AADAs at Month 36
	Age category (years)					
	2–7 (n=17)	8–14 (n=36)	15–24 (n=34)	25–65 (n=27)	Total (N=114)	Total (n=41)
Age, years, mean (SD)	5.9 (1.1)	11.5 (2.1)	18.9 (3.1)	31.7 (6.7)	17.6 (9.7)	18.1 (10.5)
Sex, male, n (%)	9 (52.9)	24 (66.7)	16 (47.1)	13 (48.1)	62 (54.4)	23 (56.1)
Years since last flare-up, median (min, max)	0.3 (0.0, 2.9)	0.5 (0.0, 13.5)	0.7 (-0.1, 6.9)	0.8 (-0.1, 15.1)	0.5 (-0.1, 15.1)	0.5 (-0.1, 13.5)
Experienced ≥1 flare-up(s) in the past 12 months, n (%)	13 (76.5)	25 (69.4)	21 (61.8)	17 (63.0)	76 (66.7)	23 (56.1)
Number of flare-ups in the past 12 months, mean (SD)	2.2 (2.6)	4.8 (9.8)	1.4 (1.8)	1.2 (1.6)	2.5 (5.9)	2.7 (6.1)

CONCLUSIONS

- As their disease progresses, it is expected that patients with FOP will have a greater need for AADAs and that their needs will change over time.
- Specific types of AADAs used at Baseline varied across age groups in this NHS.
- The new-onset use of AADAs, as observed here between Baseline and Month 36, may provide a real-world indicator of FOP disease progression, worsening mobility and functional impairment over time.

Figure 1. Use of AADAs by category



[a]The reported use of work environment adaptations by these age groups may imply that the questionnaire was completed erroneously for these participants. Percentages are based on the number of participants with non-missing data at each relevant visit. New-onset AADAs are defined as those used at the specified visit that were not used or not applicable at Baseline.

Abbreviations AADAs: aids, assistive devices and adaptations; ACVR1: activin A receptor type I; FOP: fibrodysplasia ossificans progressiva; HO: heterotopic ossification; NHS: natural history study; SD: standard deviation.

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