Achondroplasia Caregiver Survey – A global perspective on diagnostic pathways, healthcare management and personal impact from carers of children with achondroplasia

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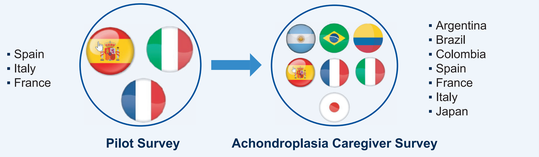
Background and Aims

- Achondroplasia is the most common skeletal dysplasia, occurring in 1/25,000 births[1]
- It is characterized by disproportionate short stature and is associated with multiple serious complications which can impact both functional and psychosocial quality of life[2]
- Among parents/caregivers (carers) of children with achondroplasia, we aimed to:
  - Elucidate the diagnostic pathway and pathways of care (including types of plans, level of support) post-diagnosis
  - Identify the primary physician responsible for care, other specialities involved and the frequency of visits
  - Understand the social/psychological support received by carers

Methodology

- Feedback from a pilot survey conducted in Europe (Spain, Italy, France) between November 22, 2018 to January 31, 2019 was used to refine the Achondroplasia Caregiver Survey
- Survey comprised questions about demographics, diagnosis, healthcare management, and provision of information and psychosocial support
- Surveys were translated to local language and sent from patient advocacy groups to their members
  - In Europe (EU) and Latin America (LATAM), carers completed an online survey between June 21 and August 21, 2019
  - In Japan, carers completed a print survey between August 30 and October 30, 2019
    - In the Japanese version of the survey, questions probing primary physician were omitted because pediatric endocrinologists always serve in this role (due to growth hormone being standard of care in Japan)



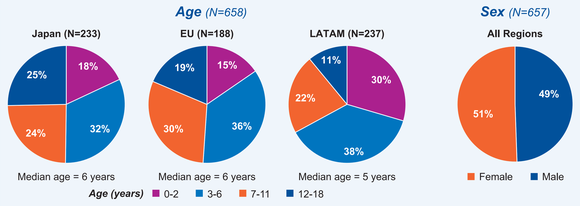
- Inclusion criteria: Parent, guardian or caregiver of a child under 18 years of age with a diagnosis of achondroplasia
- All analyses are descriptive and the number responding to each question is presented

Results

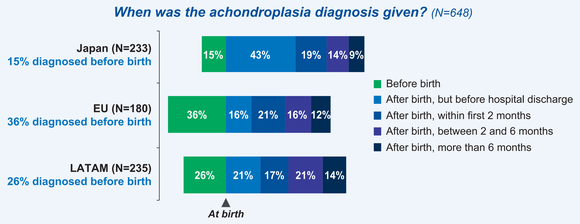
Participant Demographics

- 660 carers completed the survey
  - Japan: N=233; EU: N=190 [Spain: n=128; France: n=40; Italy: n=22]; LATAM: N=237 [Brazil: n=170; Argentina: n=36; Colombia: n=31]

Characteristics of Children Being Cared for by Participants

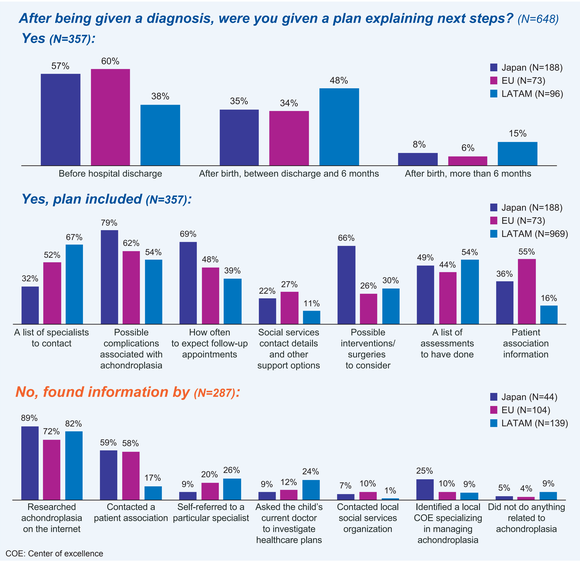


Achondroplasia Diagnosis

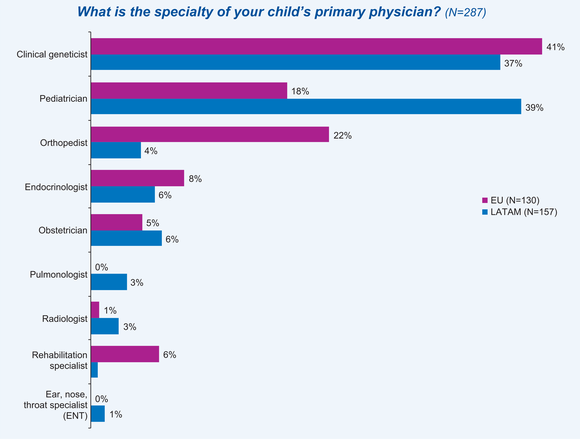


- Timing of and specialists involved in diagnosis varied considerably by region; most common specialists were clinical geneticists, pediatricians, endocrinologists and obstetricians

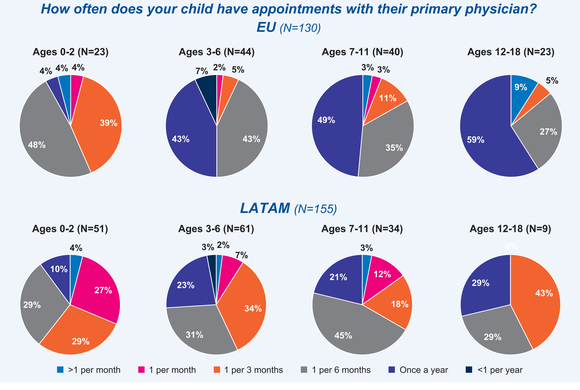
Achondroplasia Management Plan



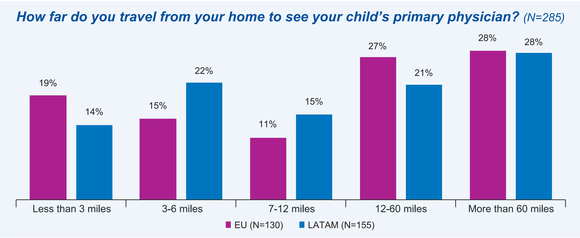
Primary Physician



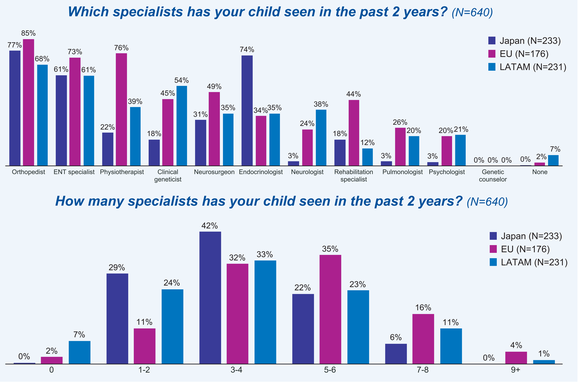
Appointment Schedule by Age



Distance Traveled to Appointments

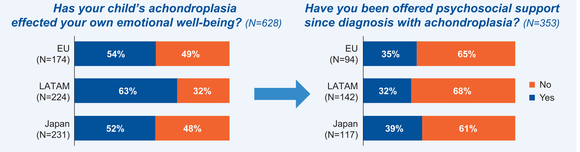


Other Specialists Seen by Patients with Achondroplasia



- In EU and LATAM, older age groups had fewer visits

Caregiver Well-Being and Psychosocial Support



Conclusions

- Achondroplasia management can be complex, requiring coordination among numerous different healthcare specialties
- There is substantial healthcare resource use among children with achondroplasia
  - Children are seeing multiple other specialists in addition to their primary physicians for repeated appointments and often have to travel substantial distances
- There are inconsistencies across countries/regions in many aspects of achondroplasia management, including having a primary physician, following a healthcare management plan, and provision of information and social/psychological support
- Improved coordination of care could potentially lessen the care burden on families
- Development of consensus-based management guidelines for achondroplasia could help improve consistency of care globally

References

1. Foreman et al. Birth prevalence of achondroplasia: A systematic literature review and meta-analysis. Am J Med Genet. 2020 Oct;182(10):2297-2316. **2.** Hoover-Fong J et al. Lifetime Impact of Achondroplasia: Current Evidence and Perspectives on the Natural History. Bone 2021 Feb 2;115872.

Acknowledgments

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