Poster no: C-0066

The Achondroplasia Roadmap

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Introduction

- Achondroplasia is a rare genetic condition, with a prevalence of 4.6 per 100,000, that affects bone growth and results in disproportionate short stature, short limbs, and macrocephaly[1,2]
- In addition to the associated medical complications and functional consequences, individuals with achondroplasia face significant psychological, emotional, and social challenges that can prevent their effective participation in society[3]
- There is an unmet need for comprehensive information for parents, healthcare professionals, and others about achondroplasia. In particular, parents lack resources to help them navigate the emotional and social aspects their children face growing up
- We have developed the Achondroplasia Roadmap, a tool primarily for parents that provides a holistic approach to explain the challenges and complexities of achondroplasia at different stages of childhood

Conclusions

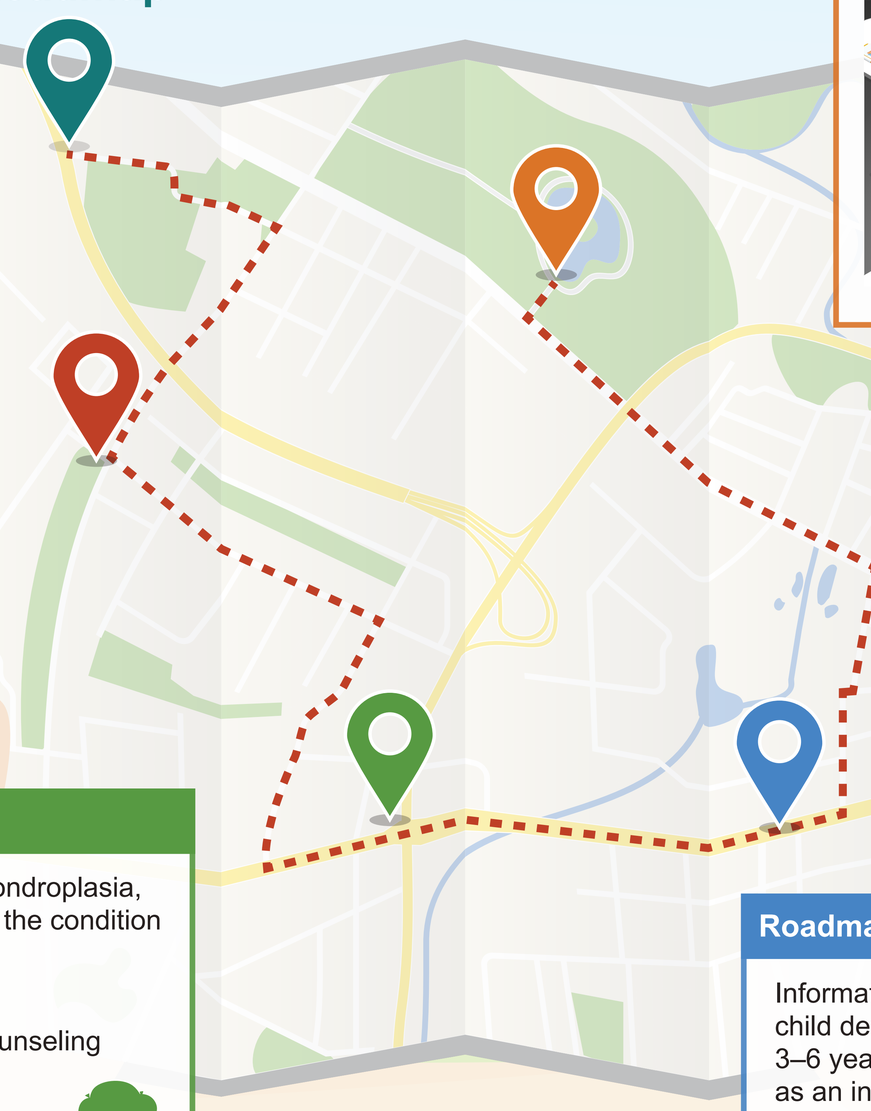
- The Achondroplasia Roadmap is a unique, comprehensive resource that is based on a person-centered approach
- Engagement with the international achondroplasia community has provided insights from diverse cultural and socio-economic backgrounds and leveraged direct experience and perspectives from those living with achondroplasia and their parents
- The resource will help raise awareness and understanding about the challenges faced by families and children with achondroplasia within medical communities as well as wider social and education networks

Methods

- International perspectives from patient associations, individuals with achondroplasia, and parents of children with the condition were collected from 11 achondroplasia patient advocacy organizations from across Europe and South America via a series of virtual meetings
- Based on guided discussions, major life milestones and accompanying medical, emotional and social issues that families and children may face were identified and developed into the Achondroplasia Roadmap
- Content was co-created and revised through the interactive and collaborative efforts of the patient organizations' representatives

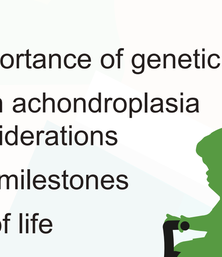
Results

The Achondroplasia Roadmap



Target audience

Primarily for parents of children with achondroplasia to help them know what to expect, find solutions, and guide and support their child in living a happy and full life

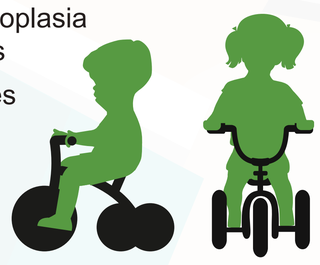


Key content

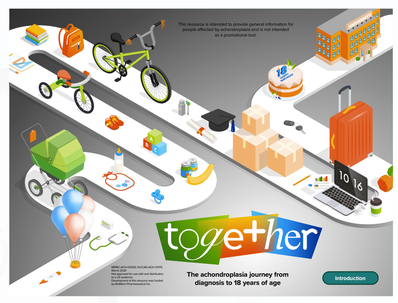
In addition to improving parents' knowledge about achondroplasia, the Achondroplasia Roadmap will help them talk about the condition with family members, teachers, peers, and others.

Key topics include:

- Diagnostic journey and the importance of genetic counseling
- Learning to care for a child with achondroplasia and managing healthcare considerations
- Navigating key developmental milestones
- Embracing the social aspects of life
- Promoting empowerment and increasing independence



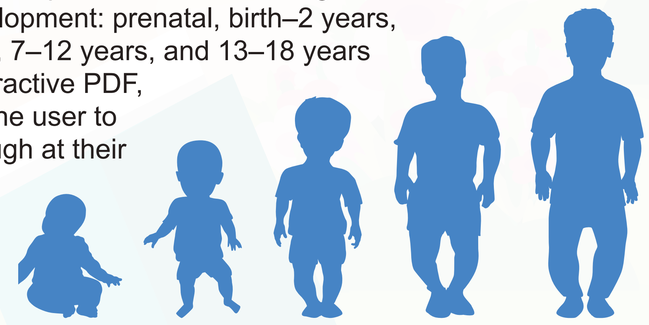
Access to the Roadmap



The Achondroplasia Roadmap is available on the websites of the partner patient organizations involved in its co-creation

Roadmap structure

Information is presented at each stage of child development: prenatal, birth–2 years, 3–6 years, 7–12 years, and 13–18 years as an interactive PDF, enabling the user to work through at their own pace



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Acknowledgments

The Roadmap was developed by the International Council of Achondroplasia Patient Association Leaders, an *ad hoc* body representing 11 countries, with support from BioMarin Pharmaceutical Inc. Medical writing assistance was provided by Maggie Lai, AMICULUM, funded by BioMarin Pharmaceutical Inc.

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Presented at the 16th International Skeletal Dysplasia Society Meeting, Madrid, Spain; September 18–21, 2024