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Patient-Centred Data Collection Provides Comprehensive Insights into Healthcare Resource Use in Achondroplasia: Data From the Pilot Phase of the VIrtual STudy in Achondroplasia (VISTA)

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Introduction

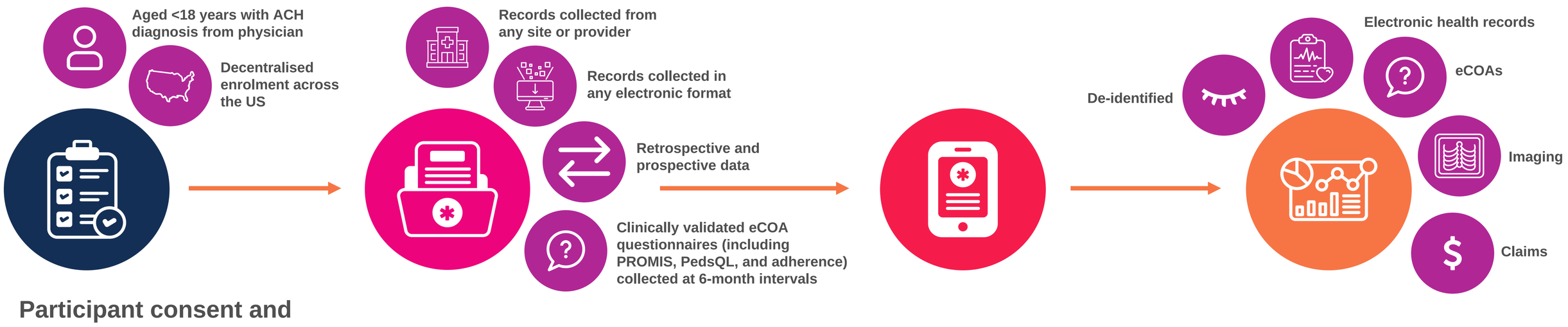
- Achondroplasia (ACH), the most common skeletal dysplasia, is caused by gain-of-function variants of the fibroblast growth factor receptor 3 (*FGFR3*) gene[1,2]
- Vosoritide, a recombinant C-type natriuretic peptide that stimulates endochondral bone growth by downregulating FGFR3 signalling, has been approved for daily dosing in the US (from birth), EU (from age ≥4 months), and elsewhere globally for children with open epiphyses[3,4]
- Whilst clinical trials of vosoritide demonstrate meaningful benefits for children with achondroplasia,[5,6] real-world data on management and treatment outcomes are limited
- The **VIrtual STudy in Achondroplasia (VISTA)** is using a participant-centred, virtual, and decentralised approach to **collect high-quality healthcare data from children with ACH in the US** and **assess the real-world effectiveness of vosoritide**

Objectives

- The objective of this pilot study was to ascertain availability of key data in real-world medical records to
  - **Evaluate the long-term impact of vosoritide treatment** on height and health outcomes in people with ACH
  - **Describe the natural history of people with ACH** in the US

Methods

The PicnicHealth digital platform enables retrospective and prospective collation of electronic health records from multiple healthcare providers and systems in the US



ACH, achondroplasia; eCOA, electronic clinical outcome assessment; PedsQL, Pediatric Quality of Life Inventory; PROMIS, Patient-Reported Outcomes Measurement Information System.

Pilot results

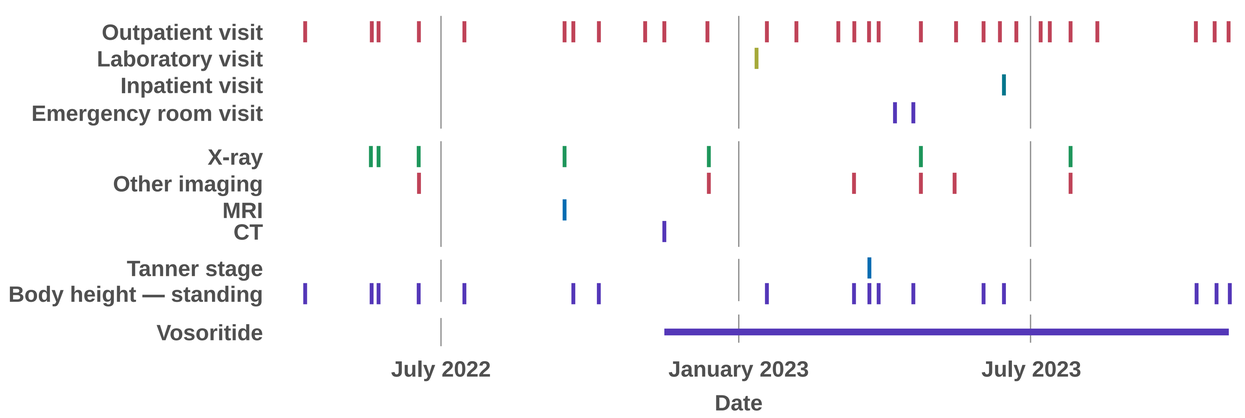
- Twenty participants have enrolled in VISTA as of 22 January 2024
  - Half (50%) were female, mainly White (70%), and 55% received vosoritide
- Participants were young when diagnosed (mean [SD] age, 1.6 [3.0] months) and at enrolment (median [IQR] age, 4.2 [1.5–7.0] years)
- The median (IQR) total duration of available clinical documentation was 4.3 (1.7–7.5) years

Resource intensive and multidisciplinary healthcare

- Median (IQR) number of healthcare settings attended was 8 (6–10), with 22 (17–30) different providers seen within settings
- Primary care physicians saw participants the most; median (IQR) annual visits was 5.8 (2.6–7.7)
  - Multiple specialists were common: 19 (95%), 18 (90%), and 15 (75%) of 20 participants had at least one visit with a geneticist, paediatric orthopedist, and otolaryngologist, respectively

A healthcare journey

VISTA enables a holistic view of individual healthcare journeys without the need for data entry by participants or investigators



Data are from a representative participant for illustrative purposes.
CT, computed tomography; MRI, magnetic resonance imaging.

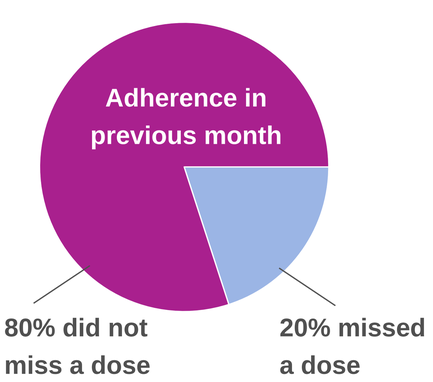
Good completion rates of clinical outcome assessments

Completion rate	PROMIS	PedsQL
Eligible children	100%	33%
Parents	67%	69%

- HRQOL questionnaires will be streamlined to improve completion rates

High vosoritide treatment adherence

- Preliminary data suggest that real-world treatment adherence was high (80%)
- Mean (SD) doses missed in past month was 3.5 (0.7)



HRQOL, health-related quality of life; IQR, interquartile range; PedsQL, Pediatric Quality of Life Inventory; PROMIS, Patient-Reported Outcomes Measurement Information System; SD, standard deviation.

Conclusions

- These data from the pilot study demonstrate the feasibility and utility of the virtual study platform for tracking the real-world, long-term impact of vosoritide on health outcomes for people with ACH
- Participants visited a variety of healthcare specialists, demonstrating the complex nature of ACH and the need for multidisciplinary care
- **VISTA is currently open for enrolment in the US**

References

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Disclosures

JMP, **DC**, **SD**, **VH**, and **FF** are current or former employees or shareholders of BioMarin Pharmaceutical Inc.
SA and **JC** are employees of PicnicHealth, who was contracted by BioMarin Pharmaceutical Inc. to perform this research.

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