

International comprehensive consensus issued on management of achondroplasia

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Fifty-five experts from 16 countries have collaborated on a new



consensus statement on the management of achondroplasia (ACH), a genetic disorder that interferes with bone growth and causes dwarfism (short stature). ACH is the most common skeletal dysplasia that is estimated to affect one in 20,000 children born worldwide.

The recommendations were published in the *Nature Reviews Endocrinology* journal, addressing the need to standardize medical, functional, and psychosocial care for people with ACH across all major life stages to improve clinical outcomes and quality of life.

Of the 55 renowned experts, five come from Nemours Children's Health, the worldwide leader for the complete medical and surgical management for children with ACH.

"The coming together of this extraordinary panel of <u>experts</u> will have a far-reaching impact on the care of patients with ACH," said Michael Bober, MD, lead author of the statement and director of the Skeletal Dysplasia Program at Nemours Children's Health. "These recommendations will help patients who otherwise wouldn't have access to the care they need and deserve. By creating these important guidelines, we hope to bridge the divide in <u>health</u> inequities for ACH patients around the globe."

The <u>consensus statement</u> addresses the entire lifespan of people born with ACH, including recommendations for diagnosis, counseling, and pregnancy of women with ACH or those carrying a baby with ACH. The recommendations also cover multidisciplinary care management for infants, children, adolescents, and adults for many common conditions that are unique to people with ACH. The statement outlines how various specialties ranging from orthopedics to otolaryngology to psychology should be mindful of the care they provide to these patients.

Among many recommendations, the authors provide guidance for



various medical sub-specialties to address common health challenges of people with ACH, including <u>spinal stenosis</u>, hearing loss, obesity, and sleep and mental health problems.

More information: Ravi Savarirayan et al, International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia, *Nature Reviews Endocrinology* (2021). DOI: 10.1038/s41574-021-00595-x

Provided by Nemours Children's Health System

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