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Advancements in achondroplasia: Clinical report provides updated guidance

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An AAP clinical report on caring for children with achondroplasia offers new information on clinical methods for monitoring potential complications and provides treatment options and resources.

Although most children with achondroplasia do well and are of normal intelligence, they often have delayed motor milestones, otitis media and bowing of the lower legs. Some health complications can occur in adulthood, so it is important to identify patients at highest risk and intervene to help prevent serious sequelae.

The report, *Health Supervision for People With Achondroplasia* from the AAP Council on Genetics and updated from 2005, is available at https://doi.org/10.1542/peds.2020-1010 and will be published in the June issue of *Pediatrics*.

Achondroplasia is the most common condition linked with severe short stature, affecting one in 10,000-30,000 children. Characteristics include rhizomelia, macrocephaly and midface hypoplasia. Some infants and children can have health consequences related to craniocervical junction compression due to a relatively small foramen magnum, hydrocephalus, upper-airway obstruction or thoracolumbar kyphosis.

Chronic pain also can begin in childhood and magnify into adulthood without intervention. Potential psychosocial problems also are a concern.

About 75%-80% of individuals with achondroplasia are born to parents with average stature, and a person with this condition has a 50% chance of passing it to offspring. While it is unnecessary to perform molecular testing in every child with a clinical diagnosis of achondroplasia, the report states that FGFR3 testing should be considered when a confirmed diagnosis is needed. Infants or children with an atypical presentation may have a *second* genetic condition and should be referred for clinical genetics evaluation.

The clinical report offers age group specific guidance, including information on counseling expectant parents. Families can benefit from anticipatory guidance and the opportunity to network with other families of children with achondroplasia.

Resources

- Little People of America Inc.
- Clinical trials for achondroplasia