

Health Supervision for People With Achondroplasia

Summary Table

This summary table—reproduced with permission from the American Academy of Pediatrics—is intended to provide support for healthcare providers to help identify individual patients at high risk of developing serious sequelae and to enable intervention before complications develop.

CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

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Health Supervision for People With Achondroplasia

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abstract

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Drs Hoover-Fong and Scott wrote this report and called a meeting from the previous AAP statement and responded to reviews. Dr. Jones provided editorial and content review. Shepherded the document through the AAP process and provided editorial and content review. All authors approved the final manuscript as submitted.

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	 Prepregnancy and Short-Stature Parents	 Prenatal and Short- and Average-Stature Parents	 Birth to 1 month	 1 month to 1 year	 1 to 5 years	 5 to 13 years	 13 to 21 years	 Adults
Physical examination	✓	✓ of fetus	✓	✓	—	—	—	—
Imaging	✓ radiographs	✓ ultrasonography of fetus	✓	—	—	—	—	—
Molecular testing	✓	✓ of fetus	✓	—	—	—	—	✓

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	 Prepregnancy and Short-Stature Parents	 Prenatal and Short- and Average-Stature Parents	 Birth to 1 month	 1 month to 1 year	 1 to 5 years	 5 to 13 years	 13 to 21 years	 Adults
Review natural history	✓ of potential offspring	✓	✓	✓	✓	✓	✓	✓
Recurrence risk and genetics	✓	✓	✓	✓	✓	✓	✓	✓
Delivery mode and location	✓	✓	—	—	—	—	—	✓
Support group(s), family support	✓	✓	✓	✓	✓	✓	✓	✓
Desired pregnancy?	—	✓	✓	—	—	—	—	✓

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		Prepregnancy and Short-Stature Parents	Prenatal and Short- and Average-Stature Parents	Birth to 1 month	1 month to 1 year	1 to 5 years	5 to 13 years	13 to 21 years	Adults
Diagnosis									
Genetic Counseling	Growth (height or length, weight, occipitofrontal circumference)	—	✓	✓	✓	✓	✓	✓	✓
	Physical examination	—	—	✓	✓	✓	✓	✓	✓
Medical Evaluation	Neurologic examination	—	—	✓	✓	✓	✓	✓	✓
	Development	—	—	✓	✓	✓	✓	—	—
Anticipation or Guidance	Neuroimaging	—	—	✓	✓ if new diagnosis	✓ as indicated	✓ as indicated	✓ as indicated	✓ as indicated
	Polysomnography	—	—	✓	✓ if new diagnosis	✓ as indicated	✓ as indicated	✓ as indicated	✓ as indicated
	Hearing assessment	—	—	✓	✓	✓	✓	✓	✓
	Radiography for kyphosis, genu varus, bowing	—	—	—	✓	✓ as indicated	✓ as indicated	✓ as indicated	✓ as indicated

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Age Group	Health Supervision for People With Achondroplasia							
	Prepregnancy and Short-Stature Parents	Prenatal and Short- and Average-Stature Parents	Birth to 1 month	1 month to 1 year	1 to 5 years	5 to 13 years	13 to 21 years	Adults
Diagnosis								
Genetic Counseling								
Medical Evaluation								
Anticipation or Guidance								
								
Warning signs of severe complications	—	—	✓	✓	✓	✓	✓	✓
Car Seats	—	✓ for hospital discharge	✓	✓	✓	✓	—	—
Achondroplasia-specific development	—	—	✓	✓	✓	—	✓	—
Jugular bulb dehiscence warning	—	—	—	✓	✓	✓	✓	✓
Supplemental security income inclusion	—	—	—	✓	✓	✓	✓	✓
Accommodations	—	—	—	—	✓	✓	✓	✓
Obesity, exercise, diet	—	—	—	—	✓	✓	✓	✓
Driving	—	—	—	—	—	—	✓	✓
College	—	—	—	—	—	—	✓	✓
Job training	—	—	—	—	—	—	✓	✓

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