



This guide is not intended to serve as medical advice or substitute for consultation with a US health care professional. Always talk to your doctor if you have any healthcare related questions or concerns.

Medical checklist for children with achondroplasia: Birth to 5 years

You can help set your child up for success by ensuring they have the right medical visits at the right times. Your child’s care team may consist of numerous providers, including those listed in the chart below. Guidelines also recommend your child be evaluated in a skeletal dysplasia clinic as soon as possible if a diagnosis of achondroplasia is made or suspected.

This checklist can help you keep track of the visits that are recommended by medical guidelines for children under 5 years old with achondroplasia. Please keep in mind that your child might not need all the tests listed here. They might also need a test that’s not listed.

	Specialist(s)	Birth to 1 month	1 month to 1 year	1 to 5 years
Diagnosis				
Physical examination	Pediatrician referral to skeletal dysplasia clinic	X	X	—
Imaging	Pediatric orthopedic specialist/surgeon	X	—	—
Molecular testing	Genetic counselor, clinical geneticist	X	—	—
Genetic counseling	Genetic counselor, clinical geneticist	X	X	X
Medical evaluation				
Physical examination	Pediatrician, clinical geneticist, pediatric endocrinologist, skeletal dysplasia clinic	X	X	X
Growth monitoring (height or length, weight, head circumference)	Pediatrician, pediatric endocrinologist	X	X	X
Development	Pediatrician, pediatric endocrinologist	X	X	X
Neurologic examination	Pediatric neurosurgeon	X	X	X
Neuroimaging*	Pediatric neurosurgeon	X	If new diagnosis	If indicated by your child’s doctor
Overnight sleep study (polysomnography)	Pediatric pulmonologist	X	If new diagnosis	If indicated by your child’s doctor
Hearing test	Audiologist	X	X	X
X-rays for curving of the upper back and bowed legs	Pediatric orthopedic specialist/surgeon	—	X	If indicated by your child’s doctor
Discuss accommodations	Physical or occupational therapist	—	—	X

*Ask your child’s physician about the importance of performing magnetic resonance imaging (MRI) in both flexion and extension positions.

References:

1. Hoover-Fong J, et al. *Pediatrics*. 2020;145(6):e20201010. 2. Savarirayan R, et al. *Nat Rev Endocrinol*. 2022;18:173–189. 3. Cormier-Daire V, et al. *Orphanet J Rare Dis*. 2022;17:293. 4. Wright M & Irving M. *Arch Dis*. 2012;97(2):129–134. 5. Pauli R, et al. *Orphanet J Rare Dis*. 2019;14:1–49. 6. Ireland P, et al. *Appl Clin Genet*. 2014. 7:117-125. 7. Merchant N, et al. *J Clin Endocrinol Metab*. 2024. 109:e1410–e1414.