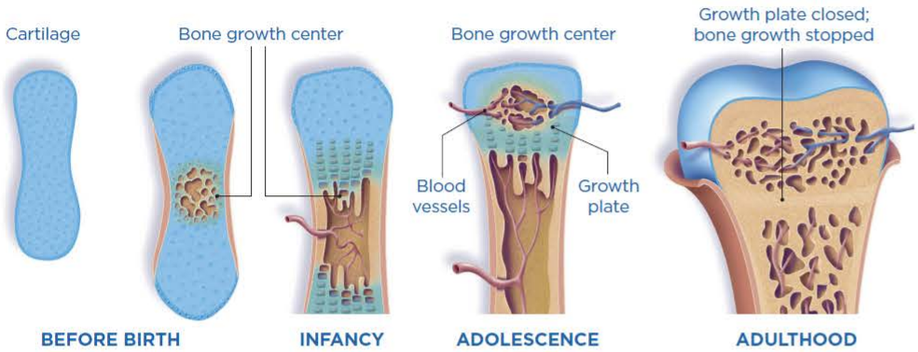
BONE GROWTH

- Soft cartilage develops into hard bone in the growth plate area, continuing until about age 16 to 20 for average stature individuals.



- Bone growth is controlled by two signaling pathways: FGFR3 which slows bone growth and NPR-B/CNP which blocks the FGFR3 signal, allowing bone growth.[1]
- In achondroplasia, growth plate activity in endochondral bone is disrupted by a mutation in the FGFR3 gene that causes it to be overactive, resulting in impaired endochondral bone development.[1]

References

1. Horton WA, Hall JG, Hecht JT. Achondroplasia. Lancet. 2007;370(9582):162-172.

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POTENTIAL COMPLICATIONS ASSOCIATED WITH ACHONDROPLASIA OVER TIME[1-5]

The complications of achondroplasia begin in infancy and are related to body-wide growth problems in each stage of physical development. Complications may build over time, and some may occur well after reaching adult stature.

INFANCY
0–1 year

- Foramen magnum stenosis
- Hydrocephalus
- Kyphosis
- Hypotonia
- Gross motor delay
- Upper airway obstruction
- Sleep apnea
- Sudden death
- Otitis media
- Hearing loss

CHILDHOOD
1–13 years

- Speech and self-care delay
- Dental malocclusion
- Lumbar hyperlordosis
- Spinal stenosis
- Genu varum

ADOLESCENCE
13–18 years

- Social adaptation challenges
- Psychosocial issues
- Obesity

ADULTHOOD
>18 years

- Chronic leg and back pain
- Cardiovascular issues
- Wheelchair bound
- Reduced life expectancy

Complications have been noted above under stage of life where they may first arise

References

1. Hunter AGW, Bankier A, Rogers JG, et al. Medical complications of achondroplasia: a multicentre patient review. J Med Genet. 1998; 35: 705-712.
2. Ireland PJ, Pacey V, Zankl A, et al. Optimal management of complications associated with achondroplasia. Appl Clin Genet. 2014;7: 117-125.
3. Hecht JT, et al. Neurologic manifestations of achondroplasia Chapter 36. Handbook of Clinical Neurology. 2014.
4. Horton WA, et al. Achondroplasia. Lancet. 2007;370: 162-172.
5. Simmons K, et al. Mortality in Babies with Achondroplasia: Revisited. Birth Defects Res A Clin Mol Teratol. 2014; 100:247–249.