intheknow
ABOUT ACHONDROPLASIA

What Is Achondroplasia?

It’s about more than just height.

Achondroplasia is a type of skeletal dysplasia (a condition that affects the bones and cartilage). While the most visible effects are in the arms, legs and face, nearly all of the bones in the body are affected. The widespread impact of this condition can cause serious, progressive and lifelong complications. But despite these complications, achondroplasia does not have to hold people back from living happy and fulfilling lives.

Achondroplasia Is

RARE

1 in 25,000 children are born with achondroplasia, and there are about 250,000 people in the world with this condition.[1,2]

GENETIC

Most children with achondroplasia (80%) are born to parents of average stature as the result of a random change in the gene (a mutation) that causes it to not function properly.[3]

DIAGNOSED IN DIFFERENT WAYS

Sometimes achondroplasia is detected or found before birth based on physical features during an ultrasound. Radiology (medical imaging) or genetic testing may be required to confirm the diagnosis. In other cases, it isn’t diagnosed until after birth.

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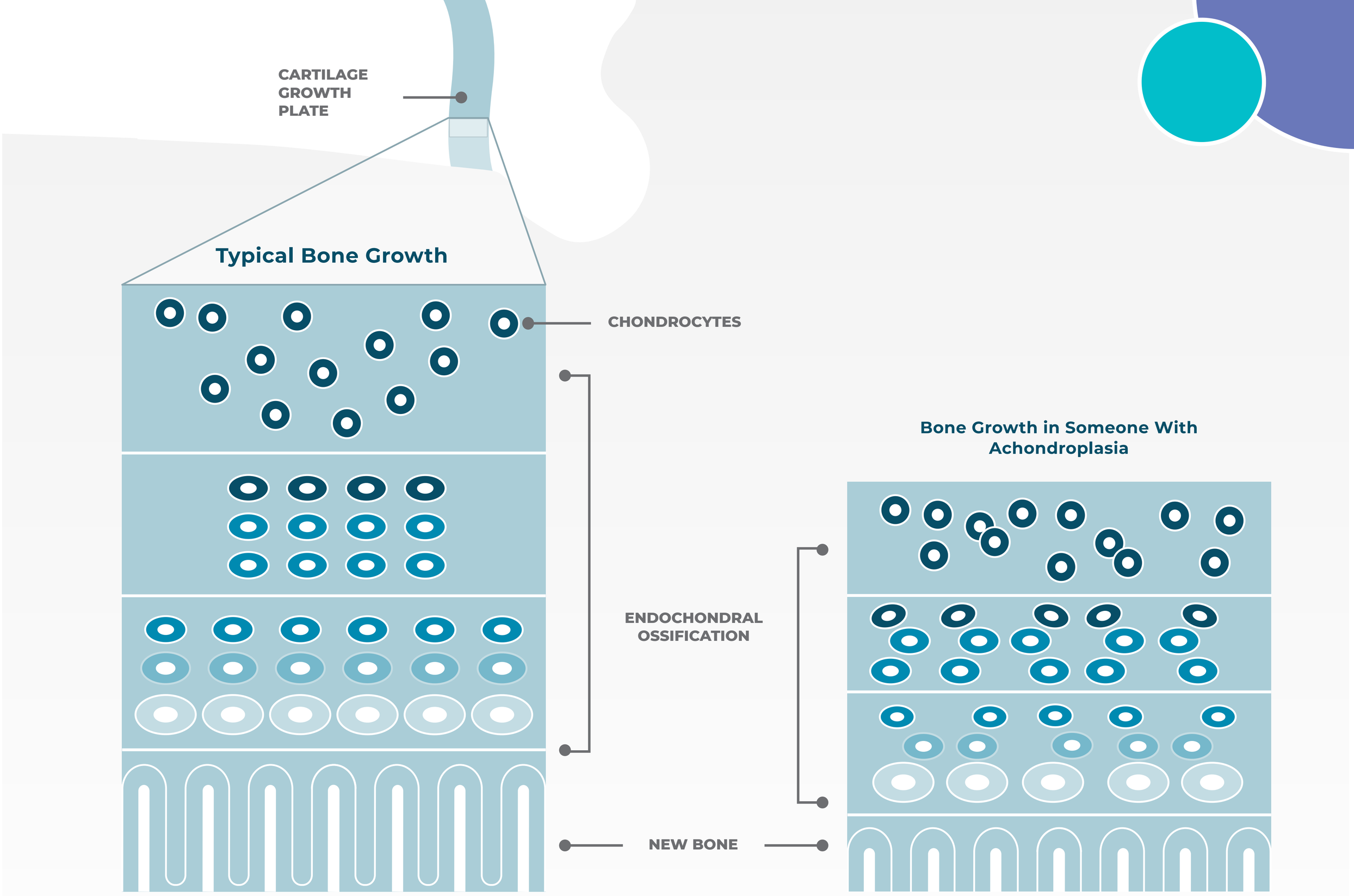
Achondroplasia Begins With the Bones

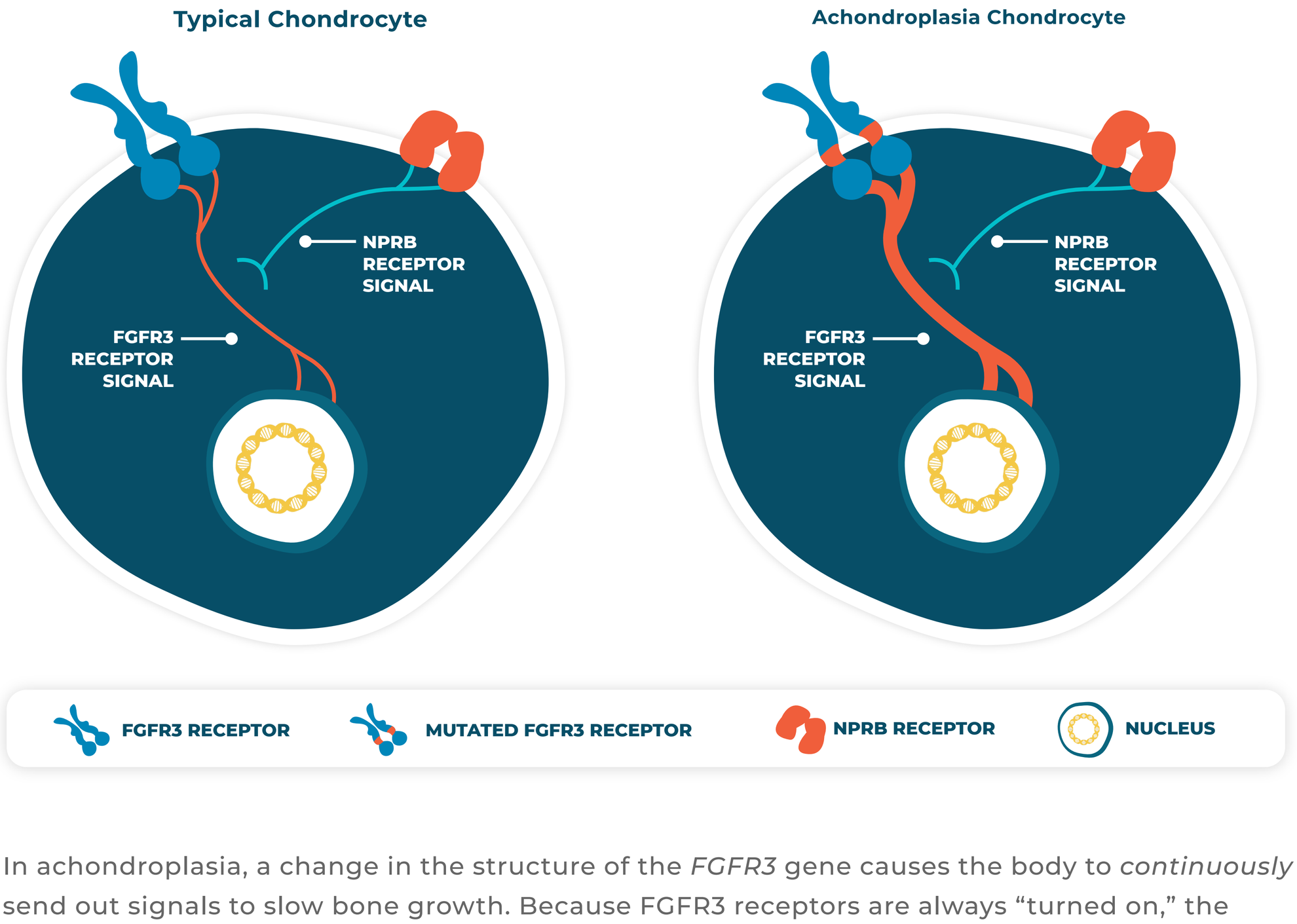
Bones begin growing before birth (in utero) and keep growing until adulthood. The process happens in the growth plates of the bones where the body makes cartilage that is then replaced by bone.

Chondrocytes (cells in the cartilage) line up to form new bone. This process is called endochondral ossification and happens in almost all of the bones in the body. Receptors in chondrocytes control the process by sending out and receiving signals.



Some signals, like the signals from NPRB receptors (natriuretic peptide receptor B), tell the bones to grow. Others, like the signals from FGFR3 receptors (fibroblast growth factor receptor 3), tell the bones to slow down growth.

FGFR3 receptors are usually only “turned on” when the body needs to stop changing cartilage into bone.



In achondroplasia, a change in the structure of the *FGFR3* gene causes the body to *continuously* send out signals to slow bone growth. Because FGFR3 receptors are always “turned on,” the signals to slow bone growth are stronger than the signals that tell bones to grow (which come from the NPRB receptors).

As a result, the chondrocytes have trouble lining up to form new bone, causing slowed bone growth.

GET IN THE KNOW

For more information about living with achondroplasia, visit:

ACHONDROPLASIA.COM

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References: 1. Ireland PJ *et al. Appl Clin Genet* 2014;7:117-125. **2.** Wynn J *et al. Am J Med Genet A* 2007;143A(21):2502-11. **3.** Hecht JT, Bodensteiner JB, Butler IJ. *Handb Clin Neurol* 2014;119:551-63.