

‘Stranger than doctors could have imagined’: Boy born without one type of brain cells

Even before he was born, it was clear that the boy’s brain was unusual—so much so that his expecting parents flew from rural Alaska to Seattle, where specialists could attend to their son from birth. That is how [pediatric geneticist] [James Bennett](#) first met the boy, then a days-old infant struggling to breathe.

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The answer was ultimately stranger than doctors could have imagined: The boy’s brain was [missing an entire type of cell, called microglia](#), the result of mutations in a single gene, called CSF1R.

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Microglia make up 10 percent of the brain’s cells, but they are not neurons and therefore have long been overlooked. The boy’s case makes their importance unmistakable. In the absence of microglia, the boy’s neurons still grew to fill his skull, but they ended up in the wrong places and made the wrong connections. Microglia, scientists have started to realize, guide the development of the brain.

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A pediatric geneticist’s job, Bennett said, is often to diagnose extremely rare conditions, which push up against the limits of the human body. “On any day, you can find a patient you spend the rest of your career thinking about,” he said. The boy is one of them.

Read full, original post: [The Boy Missing an Entire Type of Brain Cell](#)