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BIOL 294

Professor Rinehart-Kim

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**Writing Assignment 5 - Genetics in the Popular Press**

The article “Couple’s baby born without eyes due to genetic disorder less common than ‘winning the Powerball’,” by Yaron Steinbuch discusses a baby, born on November 23, 2023, who was born with haploinsufficiency of PRR12, a genetic condition that can cause a range of developmental and other abnormalities. In the baby’s case, she was born with no eyes and her eyelids were sealed shut.

At first, the baby’s parents could not figure out what was wrong as they consulted with a numerous of doctors and specialists before finally meeting Dr. Nate Jensen, a geneticist, who diagnosed their baby with a genetic mutation known as PRR12 haploinsufficiency. This condition leads to a range of neurodevelopmental challenges, which can affect various systems in the body, including the eyes. Dr. Jensen explained that this disorder is extremely uncommon, with fewer than 30 cases reported worldwide. The rarity of the condition made it difficult for the doctors to identify it quickly, and the family had to go through many medical appointments before reaching the correct diagnosis. The genetic mutation’s uniqueness and the complexity of its symptoms made it a challenging puzzle for healthcare providers to solve.

Due to this disorder being as rare as winning the Powerball, the treatment options for this particular case were limited. Since her eye sockets were empty, the doctors needed to separate them to open them in which they used spacers to ensure the baby’s facial development progressed properly. Aside from this, the baby was born healthy, and the doctors expect her to life a long and fulfilled life.

In Chowdhury’s 2023 journal article, she analyzes the symptoms and physical traits of the condition and confirms the rarity and characteristics of this genetic condition, although it may not be as odd as Dr. Jensen originally suggested. It illustrates that loss of function variants are exceptionally rare and show a high intolerance to haploinsufficiency. Similarly, the article adds that this disorder is novel and presents a broad clinical spectrum, primarily characterized by neurodevelopmental issues and eye abnormalities. While there are multiple variants, the *New York Post* article does not provide enough details to confirm which specific variant the baby has, based on the information available in both the article and the journal. Although this disorder can affect more than just the eyes, the popular article only mentions the eye-related symptoms.

Additionally, while this condition has significant physical effects on the body, it seems to be less limiting in this case. However, the journal article suggests that in other instances the disorder could lead to more severe and enervating outcomes for those affected.

Steinbuch, Yaron. Couple’s baby born without eyes due to genetic disorder less common than ‘winning the Powerball. *New York Post*. (2024). <https://nypost.com/2024/02/08/news/baby-born-without-eyes-due-to-rare-genetic-disorder/>.

Chowdhury F, et al., Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. *Genetics in Medicine: Official Journal of the American College of Medical Genetics* 23(7):1234-1245 https://doi.org/10.1038/s41436-021-01129- 6 (2021).