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Writing Assignment 5 Genetics- Related Article from the Popular Press

This article relates to genetics because it is about a baby being born with a mutation or genetic disorder called haploinsufficiency of PRR12(Proline Rich 12).

The baby was born on November 23, 2023, and when the pediatrician examined the baby, they discovered that she had no eyes. They were not underdeveloped they were not there at all. The eyelids were sealed.

Initially, the parents did not understand what was wrong with their baby girl. They took her to numerous doctors and specialists until they were able to discover from Dr. Nate Jensen who is a Geneticist that their baby suffered from a genetic mutation called haploinsufficiency of PRR12 that causes a range of neurodevelopmental issues or disorders that can affect eyes and other systems in the body. Dr. Jensen stated that this disorder is extremely rare that there are less than 30 cases at this time described in our world. Because of how rare this article states the disorder is it took a lot of doctor visits to diagnose (Steinbuch, Y. 2024)

The options or treatment for this specific incident are limited to prosthetic eyes since there is nothing in the baby's eye sockets to work with, they will have to un-fuse them to open them and provide spacers so her face will develop properly. Other than this the baby was born healthy so they expect her to live a long and happy life. They do mention that this disorder is on par with winning the lottery on the rarity.

The rarity and conditions of this mutation or disorder depending on the variant are confirmed by F. Chowdhury. However, it doesn't seem as if it is quite as rare as Dr. Jensen stated above. Also, the symptoms or physical characteristics of this disorder are confirmed in the journal article (Chowdhury F 2023). The article explains that loss of function variants is extremely rare and indicates a high intolerance of haploinsufficiency. They do advise that this disorder is a novel disorder and has a wide clinical spectrum that is marked mainly with neurodevelopmental and eye abnormalities. There are several variants, and the NY Post article does not provide enough information for a determination of the variant the baby has based on information provided in the article or from the journal entry. This disorder can affect other areas than just the eyes. The eyes were the only system mentioned in the popular article though.

While this mutation or disorder does have pretty severe physical effects on the human body it sounds like it is not as limiting as it could be, at least in this case in others from the journal article there could be more severe and limiting consequences to being born with this disorder.

References

Steinbuch, Y. (2024, 02/08/2023). Couple's baby born without eyes due to genetic disorder less common than 'winning the Powerball'. *New York Post*. 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)