Writing Assignment 5

In an article published by the *Washington Post*, written by Mark Johnson, he writes about the discovery of a new and diverse human genome called pangenome. With the discovery of this genome it has greatly shed light on rare genetic disorders that had previously fallen short on any hopes of finding out the causes of rare genetic disorders, let alone solutions for treatment. The author mentions a particular young girl named Celia Steele who is suffering from a rare genetic disorder.. Celia is a 10 year-old girl who has never walked or talked, and has endured many hardships. It wasn't until 2020 Celia's mother would finally have an answer as to what was causing her daughter's rare disorder; doctors discovered the cause came from two mutations in the gene PDE2A. It is one of the many 20,000 genes that exist in the body. This mutation is so rare that just four people in the world share her condition. What is shocking to Celia's parents is that Ceila has a twin brother who was not affected by the same mutation; her brother is perfectly healthy and normal.

In the article the author references that this genome evidence can be found in the journal Nature. The article the author is referring to can be found here at this link: https://www.nature.com/articles/s41586-023-05896-x#Sec17. What is incredible is the way that the pangenome was obtained; it was discovered by accident! During another genetic related study a full genetic blueprint with 47 different ethnicities such as Asian, American, Caribbean, European, and African were all found and their respective blueprints were recorded between 2008-2015. For the longest time, the very first draft of the human genome came from one single man who was from Buffalo. 70 percent of his genetic code has been used as reference since its release in 2001. With this crucial find, scientists are pushing to expand the new genome to where they can incorporate a diverse blueprint from around the world. The one human genome that has been used as the standard is but a small part of what's important; as Mark Johnson states, "the pangenome is a subway map, converging in parts of the sequence that are common to most people and branching out in areas where we differ" This emerging genome has some promising future application and is hoped to become more affordable.

It is because of this newly found genome that families who are affected by rare genetic disorders can have some new found hope to help understand why these rare mutations are occurring. It is even more of a great discovery in the fact that it has already helped many to obtain promising treatments.

Liao, W.-W. *et al.* A draft human pangenome reference. *Nature* **617**, 312–324 (2023).

Johnson, M. A new, more diverse human genome offers hope for rare genetic diseases. *Washington Post* (2023).