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BIOL294 Writing Assignment #3

The referenced BBC article (Pope, 2026), covers a heartwarming story of a young girl, Saffie, who has Leber's Congenital Amaurosis (LCA). LCA is a rare disease, and falls under a broad branch of inherited retinal dystrophies (IRDs). LCA is an RPE65-mediated IRD, which means that it is caused by a mutation in the gene with codes for the retinal pigment epithelium 65 kDa protein (CADTH, 2021). LCA can present at a variety of ages, and unavoidably progresses to complete blindness during the first 3 decades of life.

Doctors had given Saffie the prognosis of being blind by 30. She was already suffering from loss of peripheral vision, and very poor night and low-light vision. Her parents stated that it was heavily impeding her life and ability to do normal childhood activities such as trick-or-treating. Saffie is near-sighted and has been wearing glasses since she was 2 years old, and her diagnosis of LCA was made at the age of 5 after her parents noticed her struggling with low-light environments. She underwent plenty of testing at Moorfield Eye Hospital before treatment and was then referred to the Great Ormond Street Hospital to be treated via Luxturna therapy. She had these treatments on each eye individually, as is convention for many ocular procedures, in April 2025 and September 2025. Following treatment, Saffie is now able to see in the dark when trick-or-treating, and has noticed improvements in her peripheral vision. Cases with as much improvement as Saffie's typically only happen when treated during critical periods of visual development.

Luxturna, or Voretigene Neparvovec, is a form of gene therapy. It uses an adeno-associated virus vector (AAV2) to inject a healthy RPE65 gene into the retinal pigment epithelium (RPE). Only one dose is needed per eye, and while it cannot heal existing damage in older children, it can still slow or even stop progression. Positive results of Luxturna, such as Saffie's case, paint a hopeful picture for the future of gene therapy in treatment of inherited ophthalmic diseases. Many uncurable diseases stem from similar mutations, such as retinitis pigmentosa. While adults living with this disease may not directly benefit from these advancements, their descendants may not suffer from the same disease despite inheriting it.

The BBC news article provides accurate information both about the disease Saffie inherited, and the treatment she underwent. Referencing the CADTH (2021) review, the "blind by the age of 30" prognosis lines up well with clinical data. The symptoms reported are expected of her disease at her age. The reported results of treatment, method of treatment, and remarks regarding the realistic outcomes of treatment at various age ranges are accurate as well.

References

- Pope, Alex. (2026, April 22nd). *Girl, 6, has sight restored through gene therapy*. BBC News. https://www.bbc.com/news/articles/ce35x8759zzo
- Canadian Agency for Drugs and Technologies in Health. (2021, January). *Clinical review report: Voretigene neparvovec (Luxturna) (Novartis Pharmaceuticals Canada Inc.): Indication: Vision loss, inherited retinal dystrophy*. https://www.ncbi.nlm.nih.gov/books/NBK569039/