Library Research Award Essay

Group Project

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Rare diseases affect an estimated 1 in 10 Americans; however, there are significant barriers in rare disease research leading to treatment and therapies. Many of these diseases are so rare that only a handful of scientists worldwide are conducting research, and pharmaceutical companies lack financial incentive to produce treatments given that there is such a small target audience. To learn more about this topic, we enrolled in a class entitled *Clinical Research in Rare and Neglected Diseases* with Barbara Calhoun. Through this class, we learned about the National Organization for Rare Disorders (NORD), whose website has reports containing critical information for patient families for over 7,000 rare disorders which affect 25-30 million Americans. Unfortunately, these reports are seldom updated with current research. We discovered that the NORD report for Ellis-Van Creveld syndrome had not been updated since 2012, so we took the initiative to spend the semester researching this rare disease in order to update the report for patients and families.

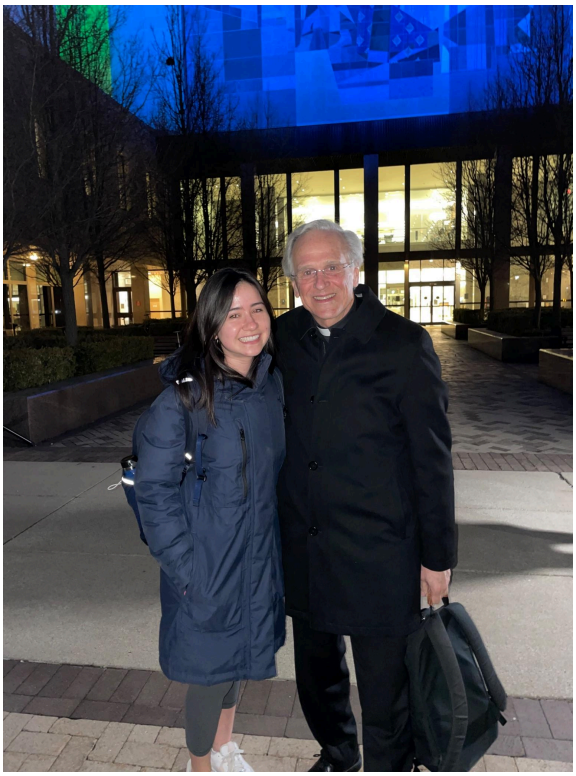
Ellis-Van Creveld syndrome (EVC) is characterized by short stature and heart defects, among a wide range of other symptoms such as extra fingers and toes. However, there are such few cases in the United States that many of the recent research papers published are in smaller, less accessible journals. Luckily, we were able to view most of these articles through access provided by the University of Notre Dame Hesburgh Libraries. We spent many hours reading and researching on OneSearch in the Hesburgh Library 2nd floor in order to conduct our literature review. We discovered that molecular testing is now available worldwide to confirm a

diagnosis of EVC, which has been documented to affect the *EVC1, EVC2, DYNC2LI1,GLI1, WDR35, PRKACA, PRKACB*, and *SMO* genes. Brooke was also able to attend the *Building a Better Lit Review with VOS Viewer* workshop by Julie Vecchio in April 2023, which was greatly helpful in visually organizing the literature review. One challenge we faced was learning how to write advanced medical discoveries at an 8th grade reading level, in order to make it concise and accessible for the general public. Throughout spring 2023, we were able to complete our literature review after many study days in the library and create a comprehensive, updated report for EVC.

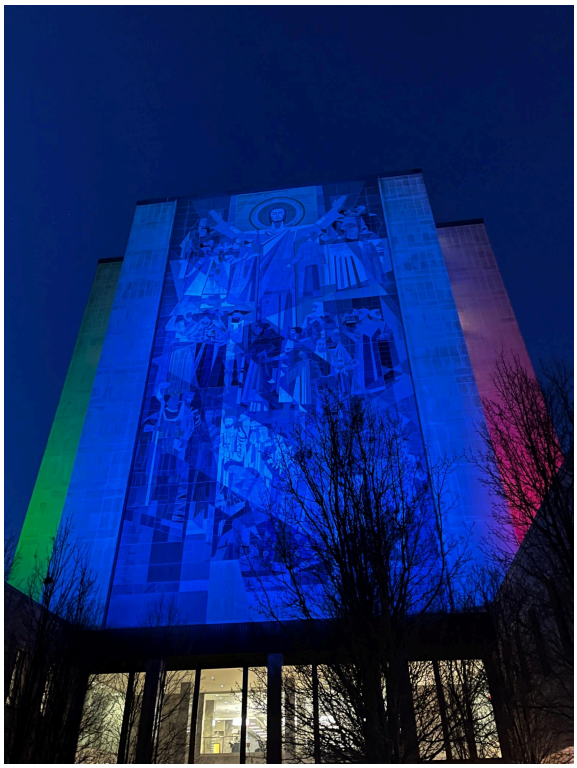
In Fall 2023, our focus turned to contacting experts to review our report for accuracy before publication. Another large challenge is that there are very few experts of Ellis-Van Creveld disease, so trying to ask for feedback on our report proved to be difficult. We spent many hours looking through research publications, with critical access provided by Hesburgh Libraries, looking specifically to find researchers who had published on EVC in the last two years. Our hope was that if they had published recently, they would be more open to assisting us with our report. After many dead ends, we were finally able to gather feedback from Dr. Marya Hameed in Pakistan and Dr. Jorge Diogo Da Silva in Portugal in early Spring 2024. At the end of Spring 2024, we were able to complete the edits, and Barbara Calhoun sent our report to the NORD administration. At present, our report has officially been published on NORD's website as of May 6th, 2024.

To further this impact on campus, we helped organize the 14th Annual Conference on Advancing Rare Disease Research, which featured talks by families and researchers across the nation. This included making luminaries with supportive messages to display in the Jordan Hall of Science, and working at tables in North and South Dining Hall to spread the message. We also

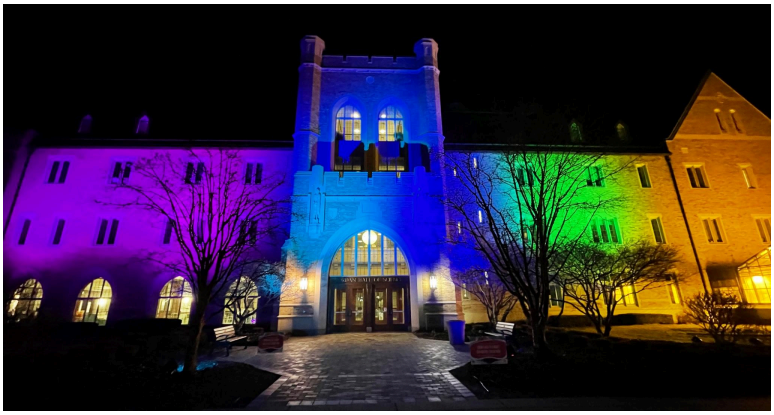
helped organize Rare Disease Day 2023 which had unprecedented attendance and included a talk by Father Jenkins. During Father Jenkins's speech, the Hesburgh Library and Jordan Hall of Science lit up using neon pink, blue, purple, and green lights, which represent the official colors of rare diseases (see photos below). It was very special to see the Hesburgh Library and The Word of Life Mural (Touchdown Jesus) lit up in honor of such an important cause.



Brooke and Fr. Jenkins at Rare Disease Day 2023



Touchdown Jesus lit in rare disease colors



Jordan Hall of Science lit in rare disease colors

While there are significant changes that need to be made to support rare disease families, our experience with NORD has shown us that we have the power to make a difference. Our Ellis-Van Creveld report on NORD's website provides updated information for patient families in a concise and accessible format. It is empowering that we are able to make a difference now, as Notre Dame students, but it is essential to continue advocating for more funding and research on rare diseases. This project would not have been possible without the assistance of the Hesburgh Library, specifically through OneSearch, the VOSViewer workshop, digital manuscript access, and reading rooms.We are truly grateful to have had this support along our journey.