Crowdfunding helps solve rare disease mystery

February 10 2015

Rare diseases—those that affect fewer than one in 200,000 people—are often identified early in life. Some 30 percent of children afflicted by these "orphan diseases" do not live to see their fifth birthday. While the U.S. Orphan Drug Act of 1983 was written into law to promote research on the topic, the cost of identifying the source and progression of these diseases remains prohibitive for many families.

But there is hope for them in our Internet age. Researchers at Tel Aviv University recently concluded a successful experiment to identify a novel genetic mutation as the source of a specific rare disease, and their experiment was supported through crowdfunding—contributions from a

large number of individuals over the Internet.

In the study, led by Dr. Noam Shomron of TAU's Sackler Faculty of Medicine and published recently in the *Journal of Genetics and Genomics*, analysis of DNA sequencing of a three-year-old girl and her family revealed a novel mutation that causes mental retardation and severe developmental delays in children. Research for the study was conducted by TAU doctoral student Ofer Isakov together with Dr. Dorit Lev and Dr. Esther Lishinsky of Wolfson Medical Center.

A roadmap to hope

"How does it help to know?" said Dr. Shomron. "It's the missing piece of the genetic puzzle, eliminating from the picture all other diseases that are known to cause death at an early age and allowing the parents to connect with families with similar problems or mutations to build a lifelong support network. The parents can connect with scientists working in the field to learn about advancements. In some cases, a change in lifestyle, drug therapy, and physiotherapy can help their child's situation. Finally, by knowing what to look for, the parents can feel free to have more children as long as they screen for the identified mutation.

"By knowing a child's DNA, you also unlock a family secret that can possibly reflect on cousins, siblings and so on. It goes around and around in a circle - who else is carrying this gene?"

The power of the crowd

"Parents look to comprehensive genetic analysis, like the one we carried out, when they don't know where else to look," said Dr. Shomron. "They are desperate to understand why their children are sick, and the medical community is challenged to identify the source of the suffering. By

travelling through the complete human genome, we are able to locate, map, and analyze mutations involved in triggering certain rare diseases.

"Crowdfunding provides the means for economically disadvantaged patients to pursue a genetic diagnosis for their ailment. Our project reached its financial goal of $5,000 within 50 days. We were pleased, to say the least. Crowdfunding is a simple and efficient solution for families with rare genetic diseases who lack private or outside funding sources."

The study harnessed whole exome sequencing (WES) to identify the genetic cause of a three-year-old girl's involuntary eye movements, small-sized head, involuntary muscle contraction, developmental delay, and progressive neurological decline. The patient had a healthy brother and there was no recent family history of neurological disorders. Although she had smiled at six weeks, laughed at three months, and reached for toys at four months, over the next two years her motor functions had degenerated and she was unable to produce words. She could neither sit nor stand unassisted and she walked only with aids. At the age of 33 months, there was no progress. Extensive medical testing produced no answers. Desperate to understand what was happening to their daughter, the parents sought answers through genetic sequencing.

But because WES costs around $1,500 per individual, and the DNA of the patient and her parents had to be sequenced as well, the price of the project came to $4,500. Due to the high expense, Dr. Shomron sought to raise the necessary funds from individual donors over the Internet. The platform they used was that of Rare Genomics Institute (RGI; http://raregenomics.org/), which specializes in this kind of fundraising in the U.S. Dr. Shomron opened the Israeli branch of RGI to cater to the local rare genetic mutation population.

Dr. Shomron is continuing his research on gene mutations to help other

families struggling with rare diseases.

Provided by Tel Aviv University