# Dr Ellie Irwin’s sight restored by pioneering genomic testing after rare eye infection



Dr Ellie Irwin, a 29-year-old doctor from Bristol, experienced a remarkable restoration of her vision just in time for her wedding earlier this year, after a five-year struggle with a debilitating and mysterious eye infection. The infection had progressed to such a severe stage that she was considering the removal of her affected eye.

Dr Irwin’s eye problems began in 2019 while she was still a medical student. Initially diagnosed with an inflammatory eye condition, her symptoms worsened despite treatment, and numerous tests designed to identify infections returned negative results. The persistent and unexplained deterioration of her vision and ongoing inflammation severely impacted her life, leading her to contemplate surgical removal of her eye.

In an effort to find an answer, Dr Irwin’s doctors at Moorfields Eye Hospital in London consulted a pioneering metagenomics team at Great Ormond Street Hospital (GOSH). This team utilised an advanced genomic sequencing technique, described as a 'genomic dragnet', capable of detecting a wide array of infections—including bacteria, viruses, and other pathogens—within patient samples without targeting a specific infection.

The metagenomics analysis identified that Dr Irwin was suffering from a rare strain of leptospirosis, a bacterial infection previously undetected by conventional tests. Upon diagnosis, Dr Irwin was prescribed a three-week course of targeted antibiotics, which led to a significant reduction in her symptoms and restoration of her sight.

Reflecting on her experience, Dr Irwin told The Independent, “I will never be able to thank the teams that continued to fight to find answers for me enough. Metagenomics has truly been game-changing for me. I spent Boxing Day of 2023 in hospital, thinking about whether it was time to have my eye removed. Now I can’t even imagine being back in that place, I am able to get back to focusing on my life—being able to have that for my wedding day is a priceless gift.”

The metagenomics testing service developed at GOSH and University College London (UCL) has been in development for over a decade and is now the first UK-accredited service of its kind. Judith Breuer, professor of virology at UCL and honorary consultant virologist at GOSH, stated, “We are now able to offer this vital genomic testing to patients around the country, and it is amazing to see the impact it is already having for patients like Ellie.”

Currently, the service processes six samples weekly from patients nationwide, typically reserved as a final diagnostic option when all other methods have failed. Carlos Pavesio, consultant ophthalmologist at Moorfields Eye Hospital, expressed his enthusiasm regarding the breakthrough: “We are delighted that this new service enabled us to identify Ellie’s infection and treat it. As a result of this, we were able to address the source of her recurrent inflammation. We are excited about the opportunities this opens up and have already initiated a clinical trial on the use of metagenomics for hard-to-diagnose eye infections.”

Dr Irwin’s case highlights the potential of cutting-edge genomic technologies to solve complex medical mysteries that conventional methods cannot, offering new hope for patients with challenging infectious diseases.

Source: [Noah Wire Services](https://www.noahwire.com)

## Bibliography

1. <https://www.gosh.nhs.uk/wards-and-departments/departments/laboratory-medicine/laboratory-database/metagenomics/> - This page details the metagenomics pathogen detection service at Great Ormond Street Hospital, which utilizes deep sequencing to identify a wide array of infections, including bacteria, viruses, and other pathogens, corroborating the advanced genomic sequencing technique described as a 'genomic dragnet' used in Dr. Irwin's case.
2. <https://www.uclh.nhs.uk/our-services/find-service/pathology-1/pathogen-genomics-unit> - The Pathogen Genomics Unit at University College London Hospitals offers access to cutting-edge pathogen genomic sequencing technologies, supporting both research and clinical applications, aligning with the genomic testing services developed at GOSH and UCL mentioned in the article.
3. <https://www.gosh.nhs.uk/our-research/our-research-infrastructure/nihr-great-ormond-street-hospital-brc/about-our-biomedical-research-centre/our-research-themes/genomic-medicine/> - This section outlines the genomic medicine theme at Great Ormond Street Hospital, focusing on using genomic technologies to support effective interventions for childhood diseases, similar to the metagenomics testing service developed at GOSH and UCL referenced in the article.
4. <https://www.ucl.ac.uk/child-health/research/genetics-and-genomic-medicine> - The Genetics and Genomic Medicine Research & Teaching Department at UCL specializes in rare genetic disorders, aiming to deliver better diagnostics and therapies, supporting the development of genomic testing services mentioned in the article.
5. <https://www.gosh.nhs.uk/our-research/a-guide-to-research-and-innovation-at-gosh/genomics/> - This page describes GOSH's leadership in genomics, delivering research and supporting care by guiding treatment, which is relevant to the metagenomics testing service developed at GOSH and UCL highlighted in the article.
6. <https://www.uclhospitals.brc.nihr.ac.uk/making-difference-rapid-pathogen-genome-sequencing-infectious-disease-diagnostics-and-surveillance> - The UCL Pathogen Genomics Unit has pioneered methods to sequence bacteria and viruses directly from clinical material, now used worldwide to monitor infections and drug resistance, supporting the advanced genomic sequencing technique used in Dr. Irwin's case.
7. <https://www.independent.co.uk/life-style/health-and-families/wedding-eye-infection-test-condition-genomics-b2741660.html> - Please view link - unable to able to access data