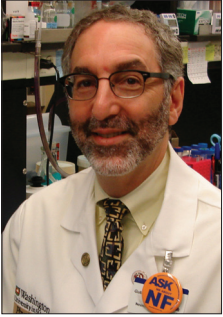


## Profile

### David Gutmann: making science work for patients



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For more on the **Washington University Neurofibromatosis Center** see <http://nfccenter.wustl.edu>

Thank goodness for academic counsellors. When David Gutmann went to see one as a 20-year-old student, he was contemplating dropping out of the University of Michigan Medical School (Ann Arbor, MI) because he found the didactic lectures dry and boring. Luckily, the counsellor persuaded him to pursue his research interests simultaneously. Gutmann enrolled in a postgraduate degree programme in human genetics, and the rest, as we say, is history: he went on to become one of the world's leading researchers and clinical experts in neurofibromatosis (NF).

Following his year studying human genetics, Gutmann took a three year leave of absence from medical school to obtain a PhD in microbiology and immunology under the mentorship of John E Niederhuber, investigating the genetic basis for immune responses in mice. He then returned to his medical training and graduated with distinction in 1986, rediscovering his passion for clinical medicine. After graduation, he completed a residency in neurology at the University of Pennsylvania (Philadelphia, PA), and then joined the laboratory of Francis S Collins as a postdoctoral research fellow. Collins and his team had just cloned the *NF1* gene, mutations in which cause NF1. Gutmann's role was to identify neurofibromin, the protein encoded by *NF1*, and investigate its function.

Initially Gutmann says his decision to work on NF was "as much chance and opportunity as it was predestination", but he then goes on to reflect that maybe his career choice was preordained. As a postgraduate student, he had been influenced by the geneticist James V Neel, who was one of the first people to discover that NF runs in families. Later, as a neurology resident, he worked with Elaine Zackai at the Children's Hospital of Philadelphia (Philadelphia, PA) in one of the largest and oldest NF clinical programmes.

In late 1993, Gutmann was recruited to the Washington University School of Medicine (St Louis, MO) to establish a clinical program at St Louis Children's Hospital. In 2004, he founded the Washington University Neurofibromatosis Center, currently composed of thirty clinical and basic science investigators. Together, they are focused on accelerating the pace of scientific discovery and its application to the care of individuals with NF.

When talking about his work at the Washington University Neurofibromatosis Center, his love of jazz comes to the fore. According to Gutmann, "two of the most important elements of jazz are improvisation and collaboration. These elements are also at the core of effective medical management for individuals with NF. As such, we require a multi-disciplinary team, composed of individuals with different expertise, but who can work

together seamlessly to address the spectrum of problems that may arise in children and adults with NF."

Gutmann's team has performed seminal research on NF1-associated optic glioma, leading to new treatments for this condition now being tested in a clinical trial (NCT01158651), but his primary goal is to one day provide personalised treatments for people with NF1. Gutmann says: "If we could predict which child is likely to develop a brain tumour and whether that optic glioma will result in vision loss, we have a unique opportunity to practise medicine in a more proactive manner. Leveraging basic laboratory investigation, we hope to one day develop patient-specific therapies for NF."

Gutmann believes firmly that the impetus for translational research should come from a desire to improve the standard of care for people affected with NF—a sentiment echoed by Daniel Geschwind at the University of California (Los Angeles, CA), who states: "Gutmann's work captures the best of translational neuroscience, in that he is a leading clinician and scientist who spends considerable time taking care of patients with the illness that he studies in the laboratory. This is true bench-to-bedside, and bedside-to-bench work."

He has received numerous accolades during his career, including the Friedrich von Recklinghausen Award—a prestigious lifetime achievement award from the Children's Tumor Foundation. However, he says that what he finds most gratifying are the interactions with his patients. Having practiced for over 20 years, some children he took care of when he first began his career are now married and have their own children with NF.

In addition to his laboratory research, Gutmann is passionate about teaching and mentoring. He is keen to spread the word about NF, which he has referred to as "the most common disorder you've never heard of". Together with his team at Washington University and St Louis Children's Hospital, he is actively developing new resources and innovative services for families with NF.

Jaishri Blakeley (Johns Hopkins University School of Medicine, Baltimore, MD), describes Gutmann as "the proverbial triple threat", referring to his research, clinical work, and mentoring and teaching work, adding he is "a living example of making science work for patients". It seems fitting, however, for the final words to come from NIH Director Francis Collins, in whose lab Gutmann trained more than 20 years ago. "David's research and clinical focus on NF1 represents a powerful merger of cutting edge neuroscience, molecular genetics, and clinical neurology."

*Sheila M Thorn*