

## 2021 Virtual NF Conference Highlights

**H**eld on June 14-16, the 2021 Virtual NF Conference brought together nearly 750 attendees from 35 countries, including members of the NF research and clinical communities, patients, advocacy groups, and industry representatives. Attendees were presented with talks given by an international group of speakers on the latest developments in research and clinical care for NF1, NF2 and schwannomatosis.

In addition to the core agenda, attendees were able to view and listen to recorded poster presentations while a panel of judges selected the top three submissions from clinical and basic science entries. The meeting wrapped up with the presentation of the 2021 Friedrich von Recklinghausen Award, given to Marco Giovannini, MD, PhD of UCLA, accompanied by tributes to Dr. Giovannini from his colleagues. Read more about the poster winners and the von Recklinghausen award on page 5.

### **NF Clinical Care Symposium**

The Clinical Care Symposium kicked off the NF Conference focusing on clinically relevant topics and projects of the CTF Clinical Care Advisory Boards (CCAB), led in the U.S. by Scott Plotkin, MD, PhD and in Europe by Pierre Wolkenstein MD, PhD and Rosalie Ferner, MD.

### **CCAB – United States**

The U.S. CCAB launched a virtual case conference in March 2021 for NF Clinic Network (NFCN) members to expand knowledge of best practices and optimize care for specific manifestations of NF. Highlights included the continued expansion of new clinics into the NF Clinic Network (now open to Canadian clinics), a publication and patient resource about the use of MEK inhibitors for NF1-associated tumors, and a survey studying the effects of COVID-19 on NF care along with research of rapid adoption of telehealth during the pandemic (with publication).

Progress has been made in the multi-year project to identify consensus guidelines for the care of NF patients, assess the level of compliance, create an educational campaign to inform clinicians, and monitor changes in guideline-concordant behavior over time. The first step was a clinician survey assessing the awareness of and agreement with the published guidelines. Results were presented by Justin Jordan, MD, MPH (Massachusetts General Hospital) at the 2020 NF Conference. The second phase of the project involved a survey within the NF Registry (May 2021). Preliminary data was presented by Vanessa Merker, PhD (Harvard Medical School) indicating that almost 15% of the 392 respondents delayed or did not get medical care in the past 12 months because of costs. Findings suggest although most patients get recommended care for many of the guidelines, there are a significant portion that had not received recommended

monitoring and surveillance or relevant cancer screenings.

### **Patient Access to Novel NF Treatments**

Laura Klesse, MD, PhD (UT Southwestern Medical Center) presented results of a survey of NFCN clinics evaluating comfort levels and identifying barriers to prescribing new NF treatments, including on-label FDA approved and off-label FDA approved medications. Approximately 93% of clinics had complete or high comfort levels of providing FDA on-label approved Koselugo (selumetinib), most often prescribed within oncology or neuro-oncology departments. The most significant impact reported was lack of familiarity with treatment options, followed by insurance barriers and patient concerns about side effects. A complementary patient survey using the NF Registry to determine patient experiences, barriers, and concerns is planned.

*Continued on page 16*

## FRIEDRICH VON RECKLINGHAUSEN AWARD WINNER

### Marco Giovannini, MD, PhD



During the virtual NF Conference the Children's Tumor Foundation announced the recipient of the 2021 Friedrich von Recklinghausen Award, Marco Giovannini MD, PhD, of the University of California, Los Angeles (UCLA). Dr. Giovannini is Professor-in-Residence at the Head and Neck Surgery at UCLA, Director of the Neural Tumor Research Laboratory, and a member of the Signal Transduction and Therapeutics Program at the UCLA Jonsson Comprehensive Cancer Center. He is also co-director of the UCLA Multidisciplinary NF Clinic.

**T**he Children's Tumor Foundation's Friedrich von Recklinghausen Award is given to individuals in the professional neurofibromatosis community who have made significant contributions to neurofibromatosis research or clinical care. It is named after Friedrich Daniel von Recklinghausen (1833-1910), the German physician who first described 'von Recklinghausen's disease' – what we now know as neurofibromatosis type 1.

As a researcher, Dr. Giovannini has had a long-term interest in the genetics and biology of human cancer predisposition and progression. His lab has made significant contributions to the characterization of many major NF tumor suppressors especially *NF2*, and *SMARCB1*. The overarching goal of his lab is to effectively translate the discoveries into medicines that will improve the quality and long-term survival of *NF2* and schwannomatosis patients. He

recognized that the lack of preclinical *NF2* and schwannomatosis models present a real roadblock to developing better treatments for patients.

Not only did his lab develop a variety of innovative cell and animal models that faithfully replicate human tumorigenesis, but he is also known to openly share the models with the community and test novel discovery paradigms swiftly.

He is creative, always open to collaborating, and is now in his third year acting as the *NF2*/Schwannomatosis Mentor to the CTF Hackathon participants. This is emblematic of his enthusiasm to share his vast knowledge, not just within the community, but outside of *NF* to help educate and raise awareness.

We are very proud and delighted to recognize Dr. Giovannini's significant achievements with the 2021 Friedrich von Recklinghausen Award. Please join us in congratulating Dr. Giovannini for this well-deserved honor.

## 2021 NF Conference Poster Session Winners

Poster sessions are an opportunity for researchers to showcase their work in the basic and clinical sciences to an audience of *NF* researchers by creating and displaying a poster that summarizes their research. A panel of judges then select the top posters, and these investigators are invited to deliver a presentation about their work in front of the full conference.

Below are the winning posters from the 2021 *NF* Conference in clinical and basic sciences.

Read the complete abstracts on the CTF newsfeed at [ctf.org/news](https://ctf.org/news).

### CLINICAL SCIENCE

- 1. Kimberly Marrs, Graduate Student, California State University, Sacramento**  
*Survey of Communication, Swallowing, and Hearing of Individuals with NF1: A Pilot Project*
- 2. Edgar Creus, IDIBELL, Spain**  
*A High-Throughput Screening Identifies the Combination of MK-1775 And Doxorubicin as a New Therapeutic Approach for MPNST*
- 3. Priya Chan, MD, Fellow, Children's Hospital Colorado**  
*Post-Operative use of MEK Inhibitors to Prevent Rebound Growth Following Partial Resection of Plexiform Neurofibromas*

### BASIC SCIENCE

- 1. Jennifer Patrilli Cram, PhD Student, Cincinnati Children's Hospital**  
*P2RY14 Modulates Schwann Cell Precursor Self-Renewal and Tumor Initiation in Mouse Model of Neurofibromatosis Type 1*
- 2. Myriam Mansour, PhD Student, France**  
*Exploring Mechanisms Driving Initiation and Progression of Plexiform Neurofibromas from Prss56Cre, Nf1fl/FI Mouse Model*
- 3. Garrett Draper, PhD Student, University of Minnesota**  
*Induced Pluripotent Stem Cell Derived Schwann Cells Harboring MPNST-Associated Mutations Fail to Escape Senescence In Vitro*