Tanja Gonska, MD

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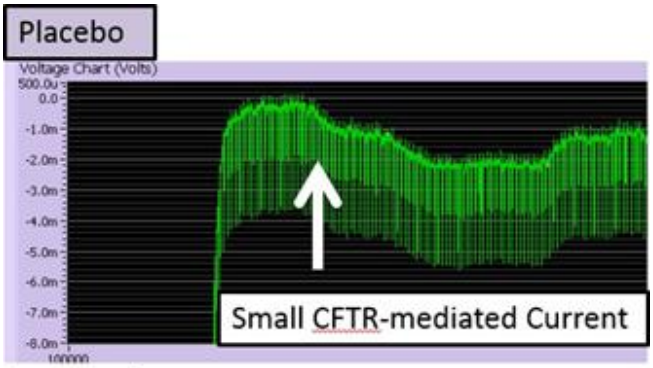
Our lab uses functional studies to study transepithelial transport in human derived tissue. We study the function of Cystic fibrosis transmembrane conductance regulator (CFTR) as well as its epithelial interaction with other ion channels and transporters that have been recently identified as modifier genes of CF disease. Further, we study the efficacy of new drug candidates in human derived tissue.

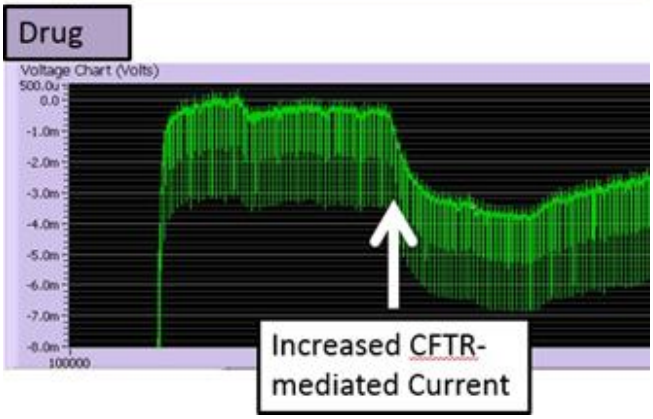
In order to understand the threshold of CFTR dysfunction that leads to organ disease or, expressed in other terms, the threshold of CFTR function that is to prevent CF organ disease, we are conducting 2 large observational clinical studies. CF-positive screened newborns that do not meet the diagnostic criteria for CF and remain in the uncertain diagnostic range due to borderline sweat tests or presence of 2 non disease-causing CFTR mutations are followed longitudinal to monitor clinically for the onset of organ disease. Similarly, adolescent and adult patients presenting with disease in CF-affected organs (acute recurrent or chronic pancreatitis, chronic sinopulmonary disease, azoospermia) who demonstrate evidence of CFTR dysfunction (borderline sweat test, borderline NPD, 2 non disease-causing CFTR mutations) are monitored longitudinally for the progression of organ disease.

CF newborn SCREENING STUDY

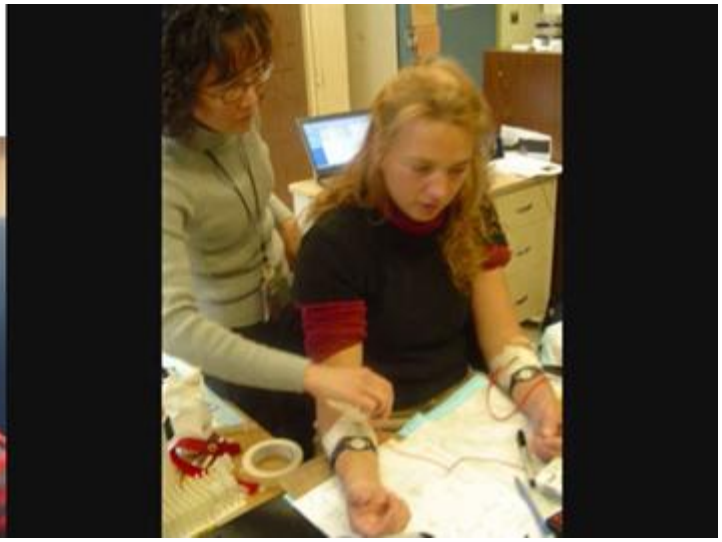
Lastly, a main area of our research activity is assigned to the development and validation of new *in vivo* measures of CFTR function that can be used as future diagnostic test and/or outcome measures in preclinical or clinical phase 2 trials.

Advancing our understanding about the level of CFTR dysfunction required for CF disease and how to measure this level of CFTR dysfunction, we are working towards novel ways for the functional assessment of the individual risk for developing CF organ disease and of the individual responses to CFTR-targeting drugs. Both tiers are essential in our journey towards personalized medicine in CF.



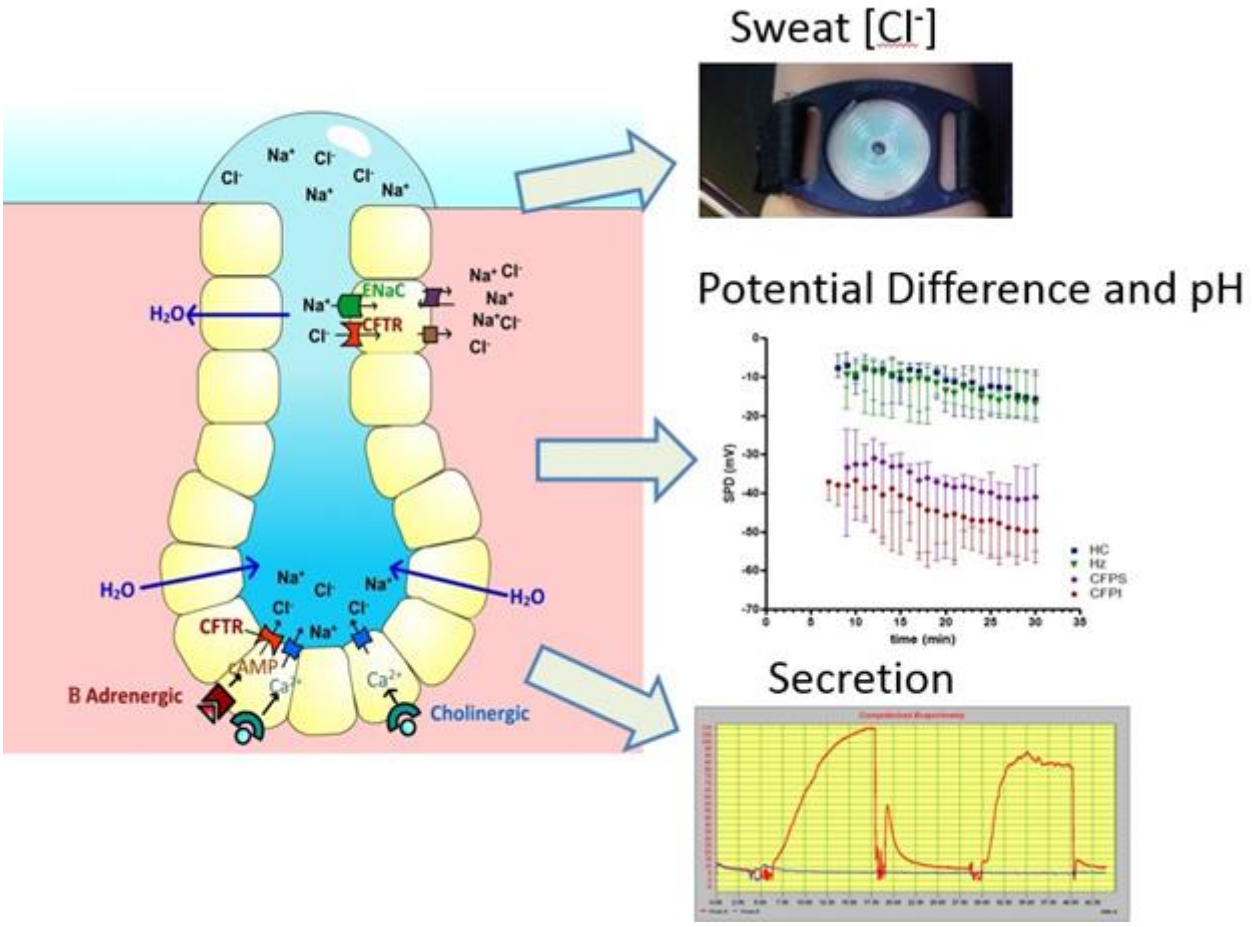


Assessing CFTR response to drugs in human tissue *ex vivo*: *Airway cells that are grown from CF lung transplants are studied to measure the effect of new drug candidates on defective CFTR chloride channel function. In future these kind of tests will help to find the best drugs or drug combination for each*



Nasal Potential Difference *(New ways to assess CFTR function in vivo): CFTR chloride channel is expressed at the lining of the airways including the nose. Its function can be measured with a small voltage sensitive catheter that is placed in the nose*.

Sweat Gland Potential Difference: *Our lab develops new tests to measure CFTR function directly in patients. One of the tests measures CFTR chloride channel in sweat glands by means of potential difference*.



New Sweat Tests: *CFTR chloride channels are abundantly expressed in sweat glands which we can now measure in different ways. These tests support making a diagnosis for CF in uncertain cases, but also to monitor response to CFTR targeting drugs in the future*. *Schematic sweat gland was designed by Stan Pasyk*.

My motivation for science and research stems from curiosity in epithelial physiological and pathophysiological processes and the fascination that functional observation can be directly tied into clinical understanding and vice versa. I am interested in research of cystic fibrosis, because to me CF is a very special disease: 1. the increasing disease complexity makes the research challenge exciting; 2. CF disease affects various organs and requires various clinical and scientific disciplines in order to understand and cure CF disease, thus there is a satisfying expertise-uniting element in CF research; and last 3. CF patients and their families that I had the privilege of taking care are especially courageous and display an endurance and motivation that is truly inspiring.

Click here for a complete list of Dr. Tanja Gonska's publications at NCBI PubMed.

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