Gaucher and Parkinson’s Recent Discoveries and Future Directions

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Collaboration
The meeting was very collaborative, and thanks to the efforts of Tony Futerman, who sets the tone of it being a “large lab meeting”, everybody was held to collaborative standards that he never wavered on. I am a big fan of this type of meeting where people with very different expertise come together to solve a single or a significant medical problem. This type of science and medical multi-discipline group works when patients are involved, or at least they are the focus of what we do. These two days of discussions had exactly that format, and I want to congratulate the CGRF on having such a terrific collaborative effort.

New Treatments
This discovery is a great opportunity for scientists and medical researchers like myself to figure out new treatments. In fact, I believe that the work on Gaucher’s disease will be an opportunity for Parkinson’s disease research and vice versa. I work very closely with the Michael J Fox Foundation for Parkinson’s Research on their scientific committees and also in their research, and they are dedicating significant funding now for studying GBA. Many scientists who have experience in both fields, including Tony Futerman and Frances Platt, can now influence and apply for funding that gives hope both to patients suffering from Parkinson’s disease or the ones at risk, as well as understanding overall the lysosomal storage diseases.

Passion for Discovery
We had many important moments and shared the passion for discovery with the families and patients present. I am certain that the research currently being conducted on Gaucher’s disease and now on a broader scale for lysosomal storage diseases, and lysosomal problems in general for neurological diseases, will lead to significant improvements in treatments. Again, I want to congratulate the Children’s Gaucher’s Research Fund and the dedicated people involved for creating such a fantastic conference, and I am eager to continue to work with the many experts in this field to make significant differences in the medical treatments of patients with Gaucher’s disease and patients at risk for Parkinson’s disease.

Parkinson’s
This was discovered a few years ago by others, and my team, which is specialized on Parkinson’s disease, realized that this was an important way to solve both disease problems in a way. The lipid storage effects on cells that create Gaucher’s disease may also have the same mechanism that causes Parkinson’s disease. It turned out not to be as simple as we first imagined, but one protein that is a problem in cells that die from Parkinson’s disease is called alpha-synuclein, and we and others have found that the more that lipids accumulate due to glucocerebrosidase (GBA) loss, the higher the risk that the cells also accumulate toxic forms of alpha-synuclein, causing degeneration of the midbrain dopamine neuron type. These are the cells, that when they die or degenerate -- are responsible for the signs and symptoms of parkinsonism.

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What did we learn?
We learned that in fact the so-called lipids (these are fat-like molecules that are normally broken down by enzymes in lysosomes—for example, by GBA. When lipids accumulate inside cells, they can create problems in nerve cells that could create Parkinson’s disease. We discussed how that occurs and relates to the fact that Parkinson’s disease usually occurs at 60-75 years of age, whereas Gaucher disease occurs in children. The reason appears to be that the very severe elevation of these so-called glycolipids is very traumatic to cells, when we are children; however, having slightly higher levels does not create any problems when we are young, but when the cells age in older brains, they also simulate the same type of problem, but not in the same types of cells as in children.

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Thanks to the compassion and energy of Deborah and Greg Macres, as well as the evolution of my scientific relationships with Tony Futerman (Weizmann Institute, Israel) and Frances Platt (Oxford University, UK), I was fortunate to be invited to this conference (sponsored by the Children’s Gaucher Research Fund) on May 6-8, 2016 at Tyson’s Corner in Virginia. It turned out to be one of the most interesting and scientifically and medically satisfying meetings I have been to for a very long time. The reason actually is the involvement of the patients and their intense focus of being able to do research that has a meaning and impact on patients’ lives, and actually also in this case, potentially the parents and grandparents. The title “Gaucher’s and Parkinson’s disease” is because when you are a parent or grandparent of a child with Gaucher’s disease, you are also carrying a gene-copy that could make it more likely to get Parkinson’s disease.

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