Dr. Berge Minassian

Dr. Minassian graduated from McGill Medical School in Montreal, trained in adult neurology at the University of California, Los Angeles and in paediatric epileptology at the Hospital for Sick Children, Toronto. Dr. Minassian spends 80 percent of his time in the laboratory to help further knowledge in his areas of specialty and design better treatments.

Rare disease are rare because they are severe. Rare disease genes therefore have a major impact on the body, and identifying these genes leads to insights into functions that are of major importance.

Dr. Minassian has focused his attention on several rare diseases. He discovered the causative genetic defects in two forms of Progressive Myoclonus Epilepsy (Lafora disease), an untreatable and fatal form of epilepsy that afflicts previously normal children and leads to their death (Nature Genetics 1998 and 2003). Dr. Minassian is now using the information from the genes to gradually understand the cellular and molecular pathways affected in these diseases. In particular, his group is uncovering a new level of control of glycogen metabolism in the brain, which when disturbed, results in hyperphosphorylation, deposition and accumulation of glycogen, which first disturbs cell function and then leads to cell death.

Dr. Minassian has also uncovered the causative defect in a form of Muscular Dystrophy, X-linked Myopathy with Excessive Autophagy. His group has shown that this disease is caused by mutations of VMA21, the chaperone that assembles the cell's acid pumps (V-ATPases). The reduction in V-ATPases results in decreased acidification of lysosomes, which leads to an arrest of autophagy, the process of cell content recycling. This results in massive accumulations of cell contents that cannot be recycled and death of skeletal muscle fibers (Cell, 2009).

Presently, Dr. Minassian's team is chasing the causative gene defect in a rare disease characterized by infantile onset Parkinsonism.

Common diseases are important because they are common. Dr. Minassian has made important contributions to common neurogenetic conditions such as Angelman Syndrome (Annals of Neurology, 1998) and Rett Syndrome (Nature Genetics 2004). He also collaborates in large studies aimed at identifying common human epilepsy genes. Finally, Dr. Minassian initiated a program to identify human epilepsy genes by first
identifying them in dogs, using saliva as a source of DNA. His group identified the first canine epilepsy gene (Science, 2005) and is now mapping several others.

Recent Publications:


