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The cause of all hereditary sensory and autonomic neuropathy type II cases has been established

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A major discovery that details the existence of a neuronal specific form of the WNK1 gene, henceforth referred to as the WNK1/HSN2 isoform, was recently completed by the research group of Dr. Guy A. Rouleau and published in the scientific journal *The Journal of Clinical Investigation*.

The group led by Dr. Rouleau is part of the University of Montreal Hospital Centre (CHUM), the CHUM Research Centre (CRCHUM) and the Sainte-Justine University Hospital Centre. Dr. Rouleau is also a professor at the University of Montreal. Their recent results will hereafter help to explain all the cases of hereditary neuropathy type II.

Hereditary sensory and autonomic neuropathy type II (HSANII) is a severe and early onset disorder that starts early during childhood. It is characterized by loss of perception to pain, touch and heat attributable to a loss of peripheral sensory nerves in the lower and upper limbs.

Unfortunately no therapy is currently available for individuals suffering from hereditary neuropathy type II. In 2004, Dr. Rouleau's team identified the DNA sequence of HSN2 as encoding a novel gene but their more recent investigations have now shown that this sequence is more precisely linked to the expression of a WNK1 isoform that is exclusively detected in the nervous system.

Till their discovery, the presence of mutations in WNK1 were only observed in individual suffering from a rare form of hypertension known as Gordon syndrome and no WNK1 isoform were exclusively expressed in the nervous system. The observation of mutations specific to the WNK1/HSN2 isoform in individuals with hereditary sensory and autonomic type II will henceforth allow the generation of animal models of the disease and help to better understand the implication of this gene and its mutations in the sensorial loss mentioned above.

Despite the severity of the symptoms observed in hereditary sensory and autonomic neuropathy type II, this important discovery points to the role of proteins like WNK1/HSN2 that are involved in the perception of pain and henceforth open new avenues for the development of pain treatments. A better understanding of the disease also provide a valuable genetic test to asses the risk or the cause of the disease in individuals at risk or presenting symptoms of it.

Source: University of Montreal