Canadian Association of Physicists | Association canadienne des physiciens et physiciennes

Contribution ID: **1789** Type: **Poster (Student, In Competition) / Affiche (Étudiant(e), inscrit à la compétition)**

POS-32 - Giant Axonal Neuropathy alters the structure of keratin intermediate filaments in human hair

Wednesday 31 May 2017 18:12 (2 minutes)

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Giant Axonal Neuropathy (GAN) follows an autosomal recessive genetic inheritance and impedes the peripheral and central nervous system due to axonal swellings that are packed with neuro-filaments. The patients display a number of phenotypes, including hypotonia, muscle weakness, decreased reflexes, ataxia, seizures, intellectual disability, pale skin, and often curled hair. We used X-ray diffraction and tensile testing to determine potential changes to the structure of keratin intermediate filaments (IFs) in the hair of patients with GAN. A statistically significant decrease of the 47 Å and the 27Å diffraction signals were observed. Tensile tests determined that the hair was slightly stiffer, stronger, and more extensible in GAN patients. These results suggest that the structure of keratin IFs in hair is altered in GAN, and the findings are compatible with an increased positional disorder of the keratin tetramers within the hair fibres. We present a detailed model how molecular disorder affects the structure of Ifs to describe our scattering data. Our results also support the use of X-ray diffraction and tensile testing as a potential diagnostic method for diseases affecting hair structure allowing for fast and non-invasive screening

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Session Classification: DPMB Poster Session | Session d'affiches DPMB (6)

Track Classification: Physics in Medicine and Biology / Physique en médecine et en biologie (DPMB-DPMB)