Abstract No. 2.2 Tuesday 5th September 2017 at 14:15-15:00

Title: To live with PHARC- a novel complex syndrome causing acquired deafblindness

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Co-presenters: Inger Marie Storaas

Main focus: Mainly practice/Acquired deafblindness

Abstract: PHARC is a newly discovered complex genetic neurological syndrome. PHARC is an acronym composed of the first letter of the five main symptoms involved in the syndrome (peripheral neuropathy, hearing loss, ataxia, retinal pigmentation, cataract) which defines it as a condition of acquired deafblindness. PHARC is very rare and there are about 50 individuals worldwide with this diagnose today. PHARC was identified as a gene mutation in 2010 by a research team at Haukeland University Hospital, Norway. Our experience is that providing information regarding consequences of PHARC to the patients and their network contributes to less social isolation and more participation for each individual with PHARC syndrome.

Our presentation will focus on the consequences of having PHARC syndrome. This will be highlighted by a personal story with a video interview with a man who has PHARC syndrome. The interview illustrates how a person with PHARC syndrome understands the initial diagnostic process and the issues regarding the physical and social consequences of living with PHARC.

PHARC is a newly discovered, complex syndrome. PHARC is very rare and there are about 50 individuals worldwide with this diagnose today. What chance do individuals with PHARC have to avoid isolation and stay socially connected?

The patients we have contact with are the first who have this diagnosis. They are the book we read, and we know little about the progression of PHARC and the future of our patients. How is it to live with such a diagnosis? What can we learn from their experiences?

Have you come across anyone with this diagnose or with similar symptoms?
If you know of a person with acquired deafblindness, showing similar symptoms, what are the possibilities for diagnosing and implementing adequate intervention procedures related to PHARC syndrome?