

ERN-RND CPMS case discussion

jour fix

Upcoming meetings:

Ataxia/HSP



Thursday, 18.11.2021, 4 – 5:30 PM CET

Cases presented:

1. Marju Herodes, Tartu, Estonia: Case of a 55-year-old-man with progressive cerebellar syndrome and with suspicion to extrapyramidal signs also. Seeking advice for diagnosis.
2. Marju Herodes, Tartu, Estonia: 51-years-old-woman with tremor, dysarthria, balance disorder, secondary parkinsonism due to antipsychotic medications (?).
3. Krista Lazdovska, Riga, Latvia: Early onset ataxia patient with WUS mutation in CCDC88C gene. Should we confirm the diagnosis of SCA 40?

Registration link: <https://us02web.zoom.us/meeting/register/tZ0vcuyrpzwrE9b58m0buKSgirhhWPuyOIJO>

Leukodystrophy



Thursday, 25.11.2021, 4 – 5 PM CET

Case presented:

Anna Zsigmond, Pécs, Hungary: Case of little girl with therapy-resistant epilepsy, severe developmental delay, progressive cerebral atrophy with the total lack of myelination, basal ganglia also affected, deceased at 3 years of age. WES was negative. Seeking final diagnosis because of family planning.

Registration link: <https://us02web.zoom.us/meeting/register/tZ0sdeCtpzgrHd32AlFwA0kdbxBxDzmmwA5MTx>

Atypical Parkinsonism



Thursday, 02.12.2021, 4 – 5:30 CET

case submission still possible!

Registration link: <https://us02web.zoom.us/meeting/register/tZcucOmpjjsiGdRXgOcG3KezI6ufLTI4pivL>

Choreas and Huntington's disease



**Friday, 17.12.2021,
10 – 11:30 CET**

case submission still possible!

Registration link: https://us02web.zoom.us/meeting/register/tZwoduqoqj4vGdRGuwz31D_Ds6HMEkNxGx_g