X-linked adrenoleukodystrophy

Digital Nordic meeting on
X-linked adrenoleukodystrophy

September 29th 2023

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Agenda

1. Introduction. Morten A Horn, Oslo University Hospital, Norway

2. Leriglitazone for children and adults with X-ALD, and brief update on allo-HSCT for adults and gene therapy cerebral ALD. Wolfgang Köhler, University of Leipzig, Germany

3. Danish natural history of X-ALD, presentation and discussion of project. Cecilie Videbæk, Rigshospitalet, Copenhagen, Denmark

4. The road ahead and structure of a Nordic collaboration on X-ALD, discussion

X-ALD – a modern view

♂

Progressive degeneration of spinal cord and peripheral nerves (myelonevropathy)

Onset typically in 20-30s

Adrenal (and testicular) failure

1/3 (?) develops fatal brain inflammation (boys mostly)

♀

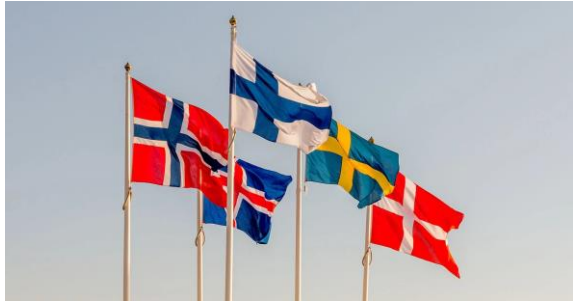
Progressive degeneration of spinal cord and peripheral nerves (myelonevropathy)

Onset typically in 40-50s

Hormonal function ~ normal

Brain inflammation very rare

A Nordic view



The Nordic countries (and the Baltics…)

- Similar size, societal structure, health care system, resources
- Traditions for collaboration and community

Possible aims for Nordic collaboration for X-ALD

- Commen strategies for diagnosis and follow-up
- Common strategies for emergent therapies; start-stop criteria
- Forum for decision-making in difficult cases
- Joint efforts in research

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Original Article

Adrenoleukodystrophy in Norway: High Rate of De Novo Mutations and Age-Dependent Penetrance

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ABSTRACT

To investigate X-linked adrenoleukodystrophy in an unselected population, we performed a population based, cross-sectional prevalence study, supplemented by a retrospective study of deceased subjects. Sixty-three subjects (34 males, 29 females) belonging to 22 kindreds were included. Thirty-nine subjects (13 males, 26 females) were alive, and 24 (21 males, 3 females) were deceased on the prevalence day. The point prevalence of X-linked adrenoleukodystrophy in Norway on July 1, 2011, was 0.8 per 100,000 inhabitants. The incidence at birth in the period 1956-1995 was 1.6 per 100,000 inhabitants. An age-dependent penetrance was observed among males and females, with more severe phenotypes appearing with rising

Topics for debate

- Should each country have a national X-ALD registry?
  - With a common minimum set of data
  - Possibility for pooling data
- Should we continue with Nordic meetings?
  - Frequency
  - Digital or physical?
- Should we have a coordinating/steering group?
  - Who would volunteer?
- Should we have a platform for difficult decisions?
  - HSCT decisions, gene therapy decisions…
  - Like for Libmeldy for MLD