

BLADDER AND BOWEL DYSFUNCTION IN X-LINKED ADRENOMYELONEUROPATHY: MALES AND FEMALES ARE AFFECTED

Hypothesis / aims of study

Adrenomyeloneuropathy (AMN) is a specific phenotype of the metabolic disorder X-linked adrenoleukodystrophy, presenting with progressive paraparesis linked to non-inflammatory degeneration of spinal tracts and peripheral nerves, as well as adrenal insufficiency¹.

Symptoms of bladder dysfunction have also been described as part of the X-ALD spectrum of disorders in an increasing collection of case reports and series¹, and represent a set of symptoms correlating strongly with reduced quality of life that are potentially treatable.

AMN is classically thought to affect males as a sex linked disorder. However it is increasingly recognised that females heterozygotic for the mutated ABCD1 gene may suffer a degree of similar symptoms to males with AMN, however without the endocrine elements of the disorder

Our study set out to catalogue the extent of urinary and bowel symptoms in a significant group of patients attending a dedicated national AMN clinic.

Study design, materials and methods

Patients attending the AMN clinic with a genetically confirmed mutation of ABCD1 were invited to participate in the cross-sectional survey. Individuals were either approached during clinic visits in person, or contacted by telephone to gain consent for participation.

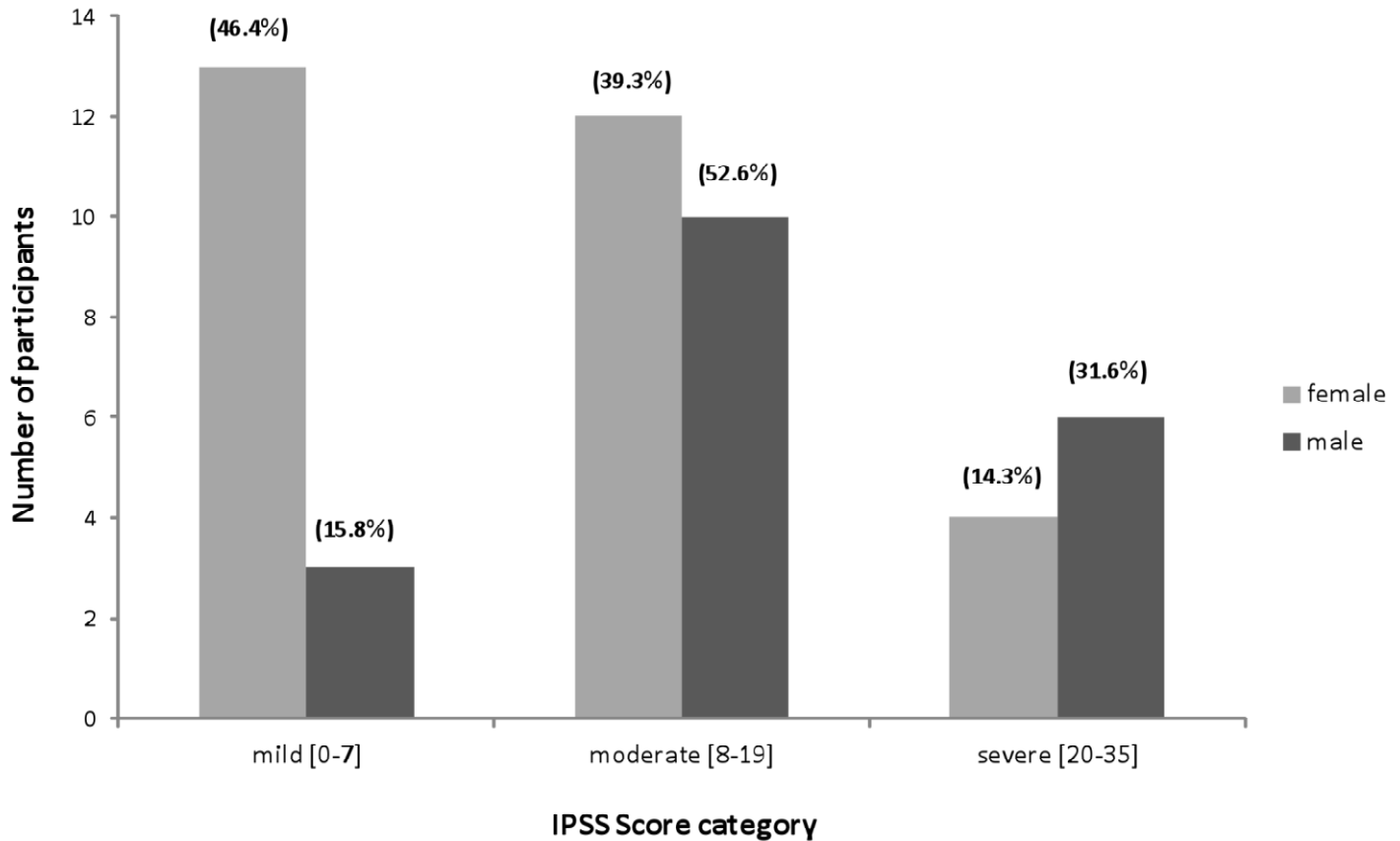
Patients were then asked to complete four validated questionnaires; the International Prostate Symptom Scale (IPSS), the Urinary Symptom Profile (USP), SF Qualiveen and the Neurogenic bowel score (NBS). These were intended to investigate overall bladder dysfunction, specific domains of bladder complaints, quality of life pertaining to urinary symptoms and overall neurogenic type bowel symptoms respectively.

Results

Of the 77 individuals registered with the AMN clinic (45 females, 32 males), 48 patients took part in interview and completed full questionnaires (29 females, 19 males), giving an overall response rate of 62.3% (64.4% of females, 59.4% of males).

The mean age for females was 46.8 years (range 20-69) and for males 47.5 years (range 21-68).

Males scored a mean IPSS score of 16.3, whilst females scored an average of 9.1 out of 35. Scores were then stratified into defined levels of severity as used for this questionnaire (mild, moderate, severe). Figure 1 shows the distribution of severity scores.



Interpretation of results

Our study suggests that male and female carriers of a mutant ABCD1 gene appear to suffer from bladder and bowel dysfunction.

Concluding message

With our study the first to investigate women specifically for bladder dysfunction, particularly in comparison to the largest male cohort to date, we have demonstrated that bladder symptoms are common in both genders.

With urinary symptoms, so common in both males and females (and to a lesser extent bowel symptoms), it should be a priority of a clinician managing patients with AMN to inquire these. This is particularly important as these are eminently treatable and can have a dramatic improvement impact on quality of life.

References

- 1) Powers JM, DeCiero DP, Ito M, Moser AB, Moser HW. Adrenomyeloneuropathy: a neuropathologic review featuring its noninflammatory myelopathy. J Neuropathol Exp Neurol. 2000 Feb;59(2):89-102.

Disclosures

Funding: This work was undertaken at UCLH/UCL Institute of Neurology and received a proportion of funding from the Department of Health's NIHR Biomedical Research Centres funding scheme. **Clinical Trial:** No **Subjects:** HUMAN **Ethics not Req'd:** It was an audit of clinical service **Helsinki:** Yes **Informed Consent:** Yes