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BLADDER DYSFUNCTION IN WOLFRAM SYNDROME IS HIGHLY PREVALENT AND PROGRESSES TO MEGCYSTIS

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Aim of the study: Wolfram syndrome is a rare genetic defect in WFS1 (Wolf syndrome type 1 - Wolframin) or WSF2(CISD2). This syndrome includes diabetes mellitus and insipidis, sensorineural deafness, optic atrophy, but not bladder dysfunction. This is, however, very common in our patients at a national referral clinic.

We aimed to quantify this problem, and test if this correlated with the genotype.

Methods: Prospective data collection for this national MDT that manages all Wolfram patients in the UK. The following data was retrospectively analyzed: date of birth, date of non-invasive urodynamics (NIU), symptoms (e.g. incontinence, urgency), bladder capacity, voided volume, post-void residual and uroflow pattern. Bladder capacity was given as % predicted bladder capacity (PBC), using (age(yrs)+1)x30(mls). Bladders were divided into normal, overactive (OAB), underactive (UAB). Symptoms, bladder behaviour and genotyping were correlated in each patient.

Data given as number (%), median (interquartile range), and analyzed by Fisher exact test or Kruskal-Wallis test, $p<0.05$ taken as significant.

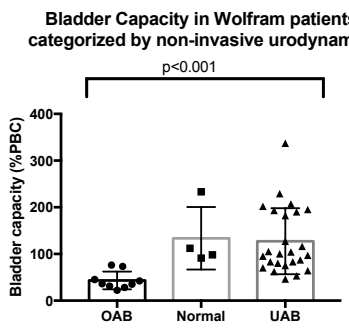
Main results: Of 40 Wolfram patients, 38 had NIU. Normal bladder function was present in 4, OAB 9, UAB 25, but symptoms were present in 11. The different patterns of bladder behaviour (OAB vs. normal vs. UAB) were significantly associated with different %PBC, 36(29.5-59)% vs. 105(93-233)% vs. 100(77.5-337)%, $p<0.001$, Figure 1a, and % emptying, 100(80-100)% vs. 100(87-100)% vs. 69(48-93)% , $p<0.05$, figure 1b.

Genotype didn't correlate with bladder behavior or symptoms.

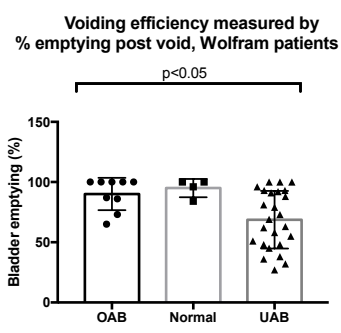
There was a progression to megacystis with time: no megacystis 13.4(9.7-16.1)yrs vs. megacystis 15.4(13.9-18.7)yrs, $p<0.05$, figure 1c.

Conclusion: This is a report of a national series of children with Wolfram Syndrome. Bladder dysfunction is very common in Wolfram syndrome (90%), but most children cope (symptoms 30%). With time there is a significant progression to megacystis, which may represent underlying neuropathic myogenic failure and is likely to require intervention in the future.

1a



1b



1c