

Urological involvement in Alstrom Syndrome

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Urinary problems seem to affect a relatively high proportion of Alstrom syndrome sufferers. During the recent international meeting, it was possible to assess 21 patients for urinary symptoms, of whom 13 were female (aged 5 – 20 years) and 8 male (8 –30 years). Broadly, the total divided equally into 3 groups of 7 – those whose urinary symptoms were either non-existent, minor or moderate- severe.

In sufferers without problems, 5 were male and 2 female. Minor symptoms affected 2 males and 5 females, and moderate – severe symptoms 1 male and 6 females.

Minor symptoms comprised long intervals between voiding (3), an urgency to void (4), others had difficulty starting to void plus poor flow (4) and lower abdominal pain (3), the latter usually associated with a need to empty the bladder.

Moderate – severe symptoms were of marked urinary frequency (5), often with urgency to void (5) and even extending to incontinence (5). Two patients had significant lower abdominal and perineal pain. Two patients have had proven highly overactive bladders which did not respond to usual drug treatments and had major surgery for urinary diversion; both were females in their late teens and both had previous difficulty in voiding. One further female of similar age has recently had difficulty voiding and has started a regime of self catheterisation to overcome this.

Three females had problems with urinary infection at ages of 2, 3 and 12 years, but these seem to have settled after appropriate treatment.

In summary, there does seem to be a definite increased risk of urinary problems in Alstrom syndrome. Although some of these are minor and require no intervention, one third of sufferers have significant symptoms which must be considered to be affecting their quality of life. Of those surveyed, the more significant problems occurred in females, 6 of 13 falling into this group versus 1 of 8 males. The most severe problems occurred in 3 females between 17 and 19 years and, interestingly, two of these seemed to change the pattern of problem from that of difficulty voiding to having overactive bladders.

These new findings suggest that Alstrom sufferers, their families and doctors should be aware of the increased incidence of urinary problems. Enquiry should be directed towards this at regular intervals, perhaps 6 – 12 monthly, and, more importantly, investigation initiated if there are suggestions of problems. The greatest problems appear to occur in females in their late teens, and an awareness of this may allow speedier diagnosis and treatment if symptoms develop, particularly as these may not be directly and obviously related to the urinary tract, such as abdominal or perineal pain. The main symptoms that may be observed are urinary frequency with urgency to void and even incontinence, but patients may also experience difficulty starting urination and a decreased flow. Some patients may have a prolonged interval between voiding, suggesting a decrease in bladder sensation. Recurring urinary

infection can also be a problem, but this will probably respond to the usual measures for this such as prolonged low dose antibiotic prophylaxis.

Investigation should start with simple measures such as testing the urine for infection, blood or protein and simple non-invasive bladder checks – urine flow rate measurement and ultrasound scanning to assess emptying. These alone may be sufficient to plan initial treatment, but if unsuccessful, further testing may involve more formal assessment of bladder function and pressures (cystometrogram) and X-rays. If the pattern of disturbance changes some of these tests may require repetition.

Treatments might usually involve drugs to regain control of an overactive bladder or learning to pass a urinary catheter to overcome voiding difficulties. Occasionally, in the most severe cases, surgical intervention may be indicated. A variety of options include using bowel patches to augment or even replace the bladder, or diverting the urine to an opening on the abdominal wall which can either be catheterised a few times each day, or simply drain into a bag. The most suitable choice requires careful discussion with your urologist, and, of course, discussion with other Alstrom families may be very helpful and rewarding.