

## **Alstrom Syndrome Scientific Meeting Morrisburg Ottawa 2001**

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### **Cardiology**

#### **Cardiac involvement:**

Recently recognised as an important feature of AS, with data from the ASI questionnaire and published case reports indicating that up to 60 % AS patients have developed heart failure due to dilated cardiomyopathy at some stage. The pattern of involvement is unusual, in that the clinical course and outcome varies according to age at presentation.

#### **Early Presentation:**

Some 35 % of AS patients experienced an abrupt onset of severe heart failure in infancy (0-6 months), usually before any visual problems manifested. Most infants survived the acute illness, with around 80% making a full recovery, which is in sharp contrast to the gloomy prognosis of other genetically determined cardiomyopathies that present at this age. Some infants did not survive, while others improved slowly or incompletely. The long-term outlook is not yet clear, as our initial optimism has been tempered by the finding that some 10 % children have relapsed. This number may increase following the introduction of routine echo assessment.

#### **Later Presentation:**

Some 25% of AS patients who were apparently unaffected in infancy develop heart failure when older (often adolescent) due to dilated cardiomyopathy with a typical progressive course.

#### **Unaffected patients:**

Around 40 % of Alstrom patients have had no reported cardiac involvement, (although not all of these have had an echo).

#### **Screening for Dilated Cardiomyopathy:**

Echo is a sensitive method of identifying dilated cardiomyopathy. There is benefit in treating patients with asymptomatic left ventricular dysfunction, so it seems logical to recommend regular echo (perhaps annual) to establish baselines, with interim echo if deterioration occurs. The signs of heart failure in children are often non-specific such as fatigue, or may be attributed to other problems eg breathlessness/asthma, so parents and clinicians must have a high index of suspicion. A note of caution - anecdotal evidence from two patients who had echo shortly before deterioration has cast doubts on our ability to predict acute events.

#### **Additional Information:**

Post-mortem histology showed severe myocardial fibrosis in two affected patients with late presentation pattern cardiomyopathy. Another patient has developed severe

pulmonary hypertension and one who was severely obese has benefited from CPAP for obstructive sleep apnoea. Pulmonary fibrosis has been identified on autopsy in one patient and may be under-recognised.