

The last international conference of Turner Syndrome Societies was held in Sydney in June 2003. Here is a small taste of the information and research provided:

Women with TS need specialised care to remain well and happy - but with proper care, their life expectancy is absolutely normal and they can live perfectly normal adult lives.

These were some of the main points made during last year's International Turner Syndrome Conference in Sydney. Speakers from all over the world converged to provide us with the latest research and information on TS.

Speakers stressed that women with TS thrive on specialised care. In fact, Dr Gerard Conway from Middlesex Hospital in London, suggested that "one-stop-shop" adult clinics should be started - perhaps around the practice of a particular doctor who is trusted in the TS community.

This doctor could invite other specialists on specific days - at first just once or twice a year. He suggested that this had many benefits, which included education and the opportunity for co-ordinated research. Women would find it far more convenient not to have to visit a number of different specialists.

Explaining the interim results from an on-going study into adult women by the National Institute of Health in the US, Dr Vladimir Bakalov said there appeared to be no weakening of the bones among TS women if oestrogen was used in adulthood.

In measuring their bone density, doctors could get a false result that appeared to indicate a thinning of bones. But measurements for TS women needed to be adjusted for smaller bones.

So far, their results indicated 43% of women with TS were lean, 35% overweight, 17% obese and that 5% were very obese. Nearly 50% had type 2 or pre diabetes. But this was not caused by excess body fat or by lack of physical inactivity.

It could be caused by a defect in insulin secretion in response to sugar, he said. But no clear answers yet existed on the long-term consequences..

Besides the well-documented heart defects associated with TS, women had been reported to have an increased risk for early coronary disease, he said. But the numbers in the US study were still too small for any firm conclusions.

However, they had found high blood pressure in 30 - 40% of TS women, high cholesterol in more than half, and an increased risk for diabetes - all risk factors for heart disease.

The family had a marked effect on behavioural prognosis for girls with TS, Mary Kaspar, from the University of New England, New South Wales, found. After studying behavioural adjustments and family, she found that fewer behavioural problems were reported for girls from families with high levels of emotional and verbal expressiveness.

Dr Charmian Quigley, paediatric endocrinologist conducted a wide-ranging toddler study in the US. Her findings showed that ear disease began early and by the age of two, over 50% needed treatment. This suggested ear disease should be looked for and treated aggressively in infancy.

By the age of two, girls were already a number of centimetres below that of the normal population. Her study was investigating starting growth hormone in infancy, so that growth never fell under the normal growth curve. She believed this could assist in the treatment of ear disease too, since these girls would show improved growth of the skull base and ear architecture.

Cognitive and behavioural interventions, she suggested, should also be started in infancy.

Dr Quigley's findings also indicated that those girls diagnosed incidentally - eg because an amniocentesis was done during pregnancy - showed significantly fewer phenotypic characteristics on average than those diagnosed traditionally.

This suggested, she stressed, that physicians should be extremely careful in advising parents on the prognosis for their daughters.

On the question of HRT, Dr Rod Baber from the Royal North Shore Hospital in Sydney said there was no doubt that the benefits outweighed risks for women with TS. Young women with TS, who had ovarian failure, should all use HRT.

After summarising all the studies so far, he said the following facts were generally agreed upon for using HRT after menopause, or after the age when menopause would naturally have occurred.

- HRT is effective in alleviating menopausal symptoms
- HRT improves bone density and reduces fractures
- HRT reduces bowel cancer incidence
- It has no beneficial effect on cardiovascular disease - at least if given orally in older women
- It increases the risk of VTE
- There is no increased risk of breast cancer if used for up to five years

- Breast cancer incidence is increased among those who use HRT for five years or more, but no increased mortality from breast cancer
- HRT can be continued after five years in lowest effective dose after discussing the individual risks and benefits

This is just a taste of the information provided by the conference. We have completed a full report-back of all papers given at the conference. If you are interested in a copy, e-mail the chairman, Jo-Anne Richards on josie@pixie.co.za.