Social life and family life in women with Turner syndrome

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• In-house family courses
• Outreach activities in users local community
• Research projects
• Information material
• Summer camps
Diagnoses

- Angelman syndrome
- Ataxia Telangiectasia
- Batten’s syndrome
- Cockayne’s syndrome and other ageing conditions
- Cornelia de Lange syndrome
- Cri du chat syndrome
- DiGeorge syndrome
- Fragile X syndrome
- Glutaric aciduria type 1
- Infantile neuronal ceroid lipofuscinosis (Seitelberger’s disease)
- Infantile neuronal ceroid lipofuscinosis (INCL)
- Klinefelter syndrome
- Leber-Weill syndrome
- Metachromatic leucodystrophy and other leucodystrophies
- Mitochondrial diseases
- Mucopolysaccharide disorders
- Muscle disorders
- Möbius syndrome
- Neurofibromatosis type 1
- Neurofibromatosis type 2
- Noonan syndrome
- Excessive growth syndromes
- Prader-Willi syndrome
- Rett syndrome
- Rubinstein-Taybi syndrome
- Soto syndrome
- Very rare chromosomal divergences, which include retardation
- Tay-Sachs disease
- Turner syndrome
- Von Hippel-Lindau syndrome
- Williams’ syndrome
- Individuals with progressive, neurological, retarded development without any known medical diagnosis

“Health status in women with Turner syndrome – a questionnaire study on health status, education, work participation and aspects of sexual functioning”

- Questionnaire sent to 168 adults over 18 yrs with TS, control group 770 women, same age group and place of residence

- Respondents:
  - 80 women in the TS group, 19-64yrs (34yrs),
  - 214 in the control group, 18-63yrs (33yrs)
Social life and family life

- Social life:
  - Education, work participation and scales from Short Form 36
- Family life:
  - Leaving parents home, cohabiting status, parenthood, aspects of sexual functioning

### Education

<table>
<thead>
<tr>
<th>Years of education</th>
<th>TS  n (%)</th>
<th>Controls  n (%)</th>
<th>P value</th>
</tr>
</thead>
<tbody>
<tr>
<td>up to 12 yrs</td>
<td>44 (55)</td>
<td>94 (44)</td>
<td></td>
</tr>
<tr>
<td>up to 15 yrs</td>
<td>23 (29)</td>
<td>77 (36)</td>
<td>0.2</td>
</tr>
<tr>
<td>&gt; 15 yrs</td>
<td>13 (16)</td>
<td>43 (20)</td>
<td></td>
</tr>
</tbody>
</table>
Studies describing educational level

- Pavlidis et al, 1995: 70% completed 4 or more years of college (aged 19-56yrs)
- Hanton et al, 2003: Mean 16 yrs of education, (aged 30-58yrs)
- F. Verlinde et al, 2004: 12.7% attended university, 7.7% in the Belgian female population, (age group 17-31 yrs)
- JC Carel et al, 2005: N=568, 322 graduated from high school or higher, 246 did not graduate from high school (age group 18-31yrs)

Work participation

<table>
<thead>
<tr>
<th>Work participation</th>
<th>TS</th>
<th>Controls</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>employed or under education</td>
<td>71 (89)</td>
<td>186 (91)</td>
<td></td>
</tr>
<tr>
<td>disability pension</td>
<td>7 (8)</td>
<td>10 (5)</td>
<td>0.096</td>
</tr>
<tr>
<td>out of work and domestic work</td>
<td>2 (2)</td>
<td>18 (5)</td>
<td></td>
</tr>
</tbody>
</table>
Work participation

<table>
<thead>
<tr>
<th>Age group</th>
<th>Working</th>
<th>Still studying</th>
<th>Disability pension</th>
<th>Unemployed</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pavlidis et al 1995</td>
<td>86%</td>
<td>5%</td>
<td>4%</td>
<td></td>
</tr>
<tr>
<td>Boman et al 2001</td>
<td>18-59 yrs</td>
<td>51%</td>
<td>37%</td>
<td>6%</td>
</tr>
<tr>
<td>Verlinde 2004</td>
<td>17-31 yrs</td>
<td>45%</td>
<td>44%</td>
<td>1%</td>
</tr>
<tr>
<td>Carel et al 2006</td>
<td>18-31 yrs</td>
<td>29%</td>
<td>50%</td>
<td></td>
</tr>
</tbody>
</table>

Short form 36, a measure for health status

- **SF36** has 8 scales, the meaning of **high scores** in 3 scales that may describe social life:

  - **Social functioning:** Performs normal social activities without interference due to physical or emotional problems
  - **Mental Health:** Feels peaceful, happy and calm all of the time.
  - **Role Functioning Emotional:** No problems with work or other daily activities as a result of emotional problems
No differences between the groups in these scales,

- But in the TS group:
  - height and level of education correlated positively to social functioning
  - age of diagnosis correlated negatively to role functioning emotional
  - hearing loss influenced all the scales

Other studies using SF 36 or other instruments

- Sylven, 1991, age>35 yrs, lived normal social lives
- Carel et al , 2006: No differences between the women with TS (18-31 yrs) and the general French population in any of the scales. All the women had been treated with GHT
- Boman 2001, Nottingham Health Profile: more social isolation than the normative group, otherwise no differences
Family life

-Leaving parents’ home,
-cohabiting status,
-parenthood,
-aspects of sexual functioning

Leaving parents’ home

- Our study, 2009: women with mean age 34 yrs, left at mean age 20.4 yrs, controls left at 18.7 yrs
- F. Verlinde, 2004: women aged mean 23 yrs, 68% still lived with their parents (39% in controls)
- K. Pavlidis et al, 1995: mean age leaving home was 22 yrs (mean age 33.8)
Cohabiting and parenthood ($p<0.001$)

<table>
<thead>
<tr>
<th></th>
<th>TS</th>
<th>Controls</th>
</tr>
</thead>
<tbody>
<tr>
<td>-live alone</td>
<td>46</td>
<td>20</td>
</tr>
<tr>
<td>-with parents</td>
<td>15</td>
<td>10</td>
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<tr>
<td>-with partner</td>
<td>39</td>
<td>70</td>
</tr>
</tbody>
</table>

- 11 women (14%) with TS had children, 110 controls (52%)

Other studies on cohabiting

<table>
<thead>
<tr>
<th>Study</th>
<th>Age group</th>
<th>Married/ living with partner</th>
<th>Single</th>
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</thead>
<tbody>
<tr>
<td>Pavlidis et al, 1995</td>
<td>19-56 years</td>
<td>45%</td>
<td>54%</td>
</tr>
<tr>
<td>Landin-Wilhelmsen et al, 2001</td>
<td>25-64 years</td>
<td>31%</td>
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<tr>
<td>Verlinde et al, 2004</td>
<td>17-31 years</td>
<td>8 %</td>
<td>14 %</td>
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</tbody>
</table>
Life Satisfaction Scale (LiSat9)

- Dark bars = Turner syndrome
- Grey bars = Control group

- P-value calculated using the $\chi^2$ statistic. Dark bars depict Turner syndrome and grey bars depict control. * $p < 0.05$

<table>
<thead>
<tr>
<th>Percentage</th>
<th>Life as a whole</th>
<th>Vocational situation</th>
<th>Financial situation</th>
<th>Leisure situation</th>
<th>Contact with friends</th>
<th>Sexual life</th>
<th>ADL</th>
<th>Family life</th>
<th>Partner relationship</th>
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<td>100</td>
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Financial and leisure situation

- Adults with TS were more satisfied with their financial situation than the control group.
- The women with TS reported more satisfaction with their leisure than controls.
Life Satisfaction Scale (LiSat9)

Dark bars = Turner syndrome
Grey bars = Control group

Conclusions

• “In spite of described delay in social and emotional development, most adult women with Turner syndrome are emotionally stable and live self-sufficient lives as independent productive adults”

E. McCauley, V. Sybert 2006