

# Social life and family life in women with Turner syndrome

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## Frambu, Norway

- In-house family courses
- Outreach activities in users local community
- Research projects
- Information
- 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
- DiGeorges syndrome
- Fragile X syndrome
- Glutarsyreuri type 1
- Infantile neuroaxonal dystrophy (Seitelberger's disease)
- Infantile neuronal ceroid lipofuscinose (INCL)
- Klinefelter syndrome
- Lesch-Nyhan 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 retarded development
- Tay-Sachs disease
- **Turner syndrome**
- Von Hippel-Lindaus 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 Form36
- Family life:
  - leaving parents home, cohabiting status, parenthood, aspects of sexual functioning



## Education

Years of education	TS n (%)	Controls n (%)	P value
-up to 12 yrs	44 (55)	94 (44)	0.2
-up to 15 yrs	23 (29)	77 (36)	
- > 15yrs	13 (16)	43 (20)	



## 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

Work participation	TS	Controls	p-value
	n (%)	n (%)	
-employed or under education	71 (89)	186 (91)	0.096
-disability pension	7 (8)	10 (5)	
-out of work and domestic work	2 (2)	18 (5)	



## Work participation

	Age group	Working	Still studying	Disability pension	Unemployed
Sylvén et al 1991	>35 yrs	96%		4%	
Pavlidis et al 1995	19-56 yrs	86%			
Boman et al 2001	18-59 yrs	51%	37%	6%	6%
Verlinde 2004	17-31 yrs	45%	44%	4%	7%
Carel et al 2006	18-31 yrs	29%	50%		21%



## 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 34yrs, left at mean age 20.4 yrs, controls left at 18.7yrs
- 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 22yrs (mean age 33.8)



## Cohabiting and parenthood (p<0.001)

	TS	Controls
	%	%
• -live alone	46	20
• -with parents	15	10
• -with partner	39	70

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



## Other studies on cohabiting

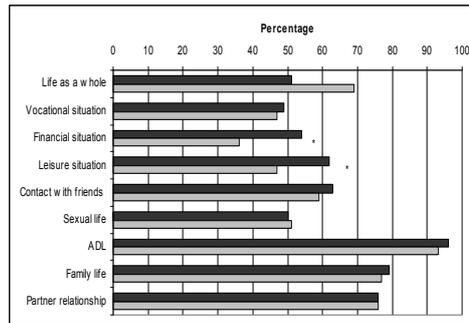


Study	Age group	Married/ living with partner	Single
Pavlidis et al, 1995	19-56 years	45%	54%
Landin-Wilhelmsen et al, 2001	25-64 years	31%	
Verlinde et al, 2004	17-31 years	8 %	14 %



## Life Satisfaction Scale (LiSat9)

Dark bars = Turner syndrome  
Grey bars = Control group



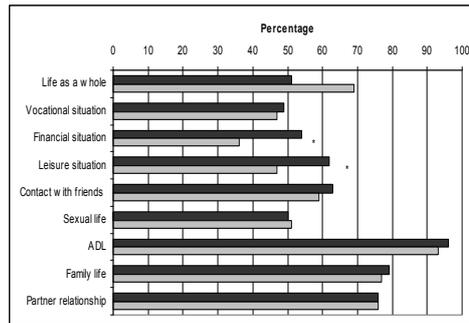
## Financial and leisure situation

- **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)

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## 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

