Assessment of medical and psychological status of women with Turner-Syndrome in young adulthood

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Background

Turner Syndrome is a clinical entity that comprises genetic, developmental, endocrine, psychosocial, reproductive and cardiovascular disorders. Turner-Syndrome patients constitute a special category of medical cases, due to the expected co-morbidities on one side, but also due to the great psycho-emotional impact of the diagnosis and treatment. Although guidelines for a functional transition do exist in the literature, some published data suggest an inadequate medical surveillance beyond transition phase, and sometimes even lost-to-follow-up. Considering the severity of some of the expected co-morbidities, along with the newly published data regarding possible fatal heart events as part of the evolution of Turner Syndrome in the early adult years, it is of great importance to assess the medical and clinical situation of former paediatric patients.

Objectives

We conducted a medical and psychological follow-up of adult patients with Turner Syndrome which had been treated at our tertiary pediatric endocrine centre.

Methods

We screened for expected comorbidities and provided a questionnaire asking for current medical care. Furthermore, we assessed quality of life with the SF36v2 forms, and general mood with the Beck-Depression Inventar.

Screening:

-height, weight, blood pressure

-QOL Analyse by means of SF-36v2 form

– Depression score using BDI-II testing

– Assessment of current medical status using questionnairs

– Biochemical analysis: hepatic and renal function, coeliac disease screening, thyroid function, vitamin D, serum estrogen

– Bone density

– Cardiac ultrasound

**Recommended cardiac-MRI and 24h blood pressure measurement*

Study population

-39 /64 contacted patients participated in the study
-Age 21 to 43 years
-23/39 45, X, 16 mosaicism
-Final height: -3,5 SD

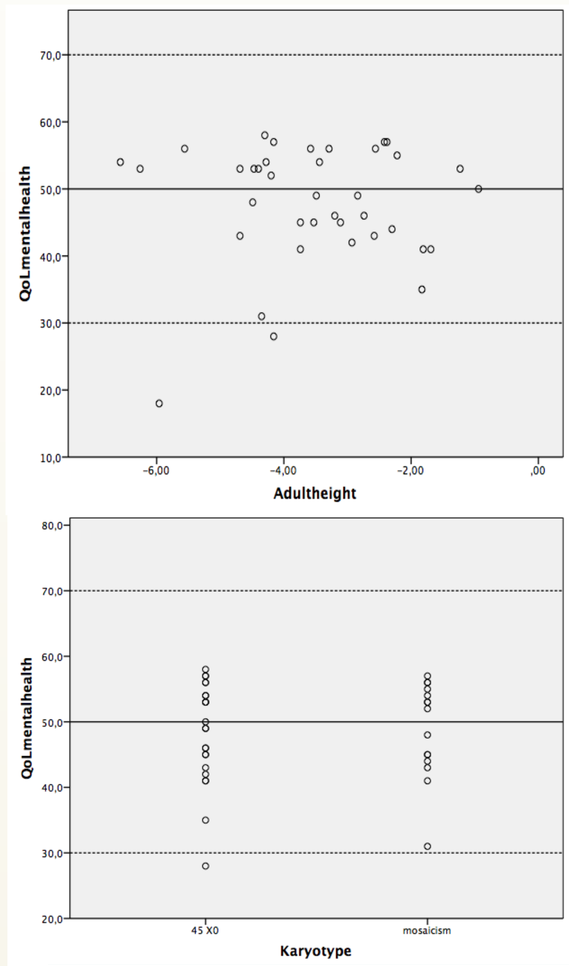
Familial status single	69,2%
High academic education	28,9%
Previous growth hormone therapy	60%
Spontaneous puberty	7,7 %
Current oestrogen substitution	81,6%
Follow-up GP	59 %
Follow-up Endocrinologist	18%
Follow-up Gynocologist	76,9%
Follow-up Cardiologist	15,4%

Comorbidities diagnosed after transition:

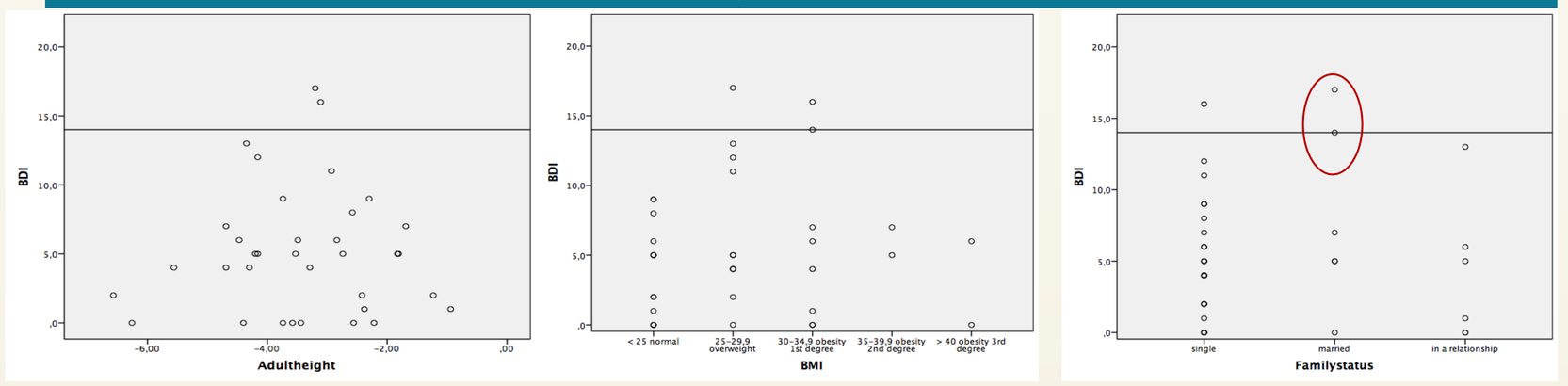
-Hypothyroidism 7, **Thyroid carcinoma: 2**
-High blood pressure: 3
-Psoriasis: 3
-Depression: 3

Results

Quality of life



BDI-II Depression Score



Scoring: 0-8 no depression, 9-19 minimal, 20-28 milde, 29-63 severe

Subjective vs. objective assessment of current medical status

Current medical care judged by patient	
Adequate	Inadequate
21	12

Current medical care judged by investigators*	
Adequate	Inadequate
5	32

**adequate: yearly clinical check-up, blood analysis, blood pressure, and every 3-5 years cardiac ultrasound, DEXA, hearing test, thyroid function, screening for coeliac disease*

Oestrogen Status subjectively	**Adequate** (no symptoms)	**Inadequate** (symptoms)
34 patients	22 (64%)	11 (36%)

Oestrogen Status objectivly	**Adequate E2 Spiegel >60ng/l**	**Inadequate**
39 patients	17 (43,5%)	22 (56,5%)

Disscusion points

-good QoL scores despite short stature and comorbidities
-standardized assessment of QoL
-low depression rate
-newly diagnosed comorbidities during this study
-70% would favour a combined care (GP and specialist)
-70% say they don‘t require psychological support

Conclusions

The surprising results might indicate a tendency to minimalize the symptoms and underestimate the importance of regular follow-up in adult Turner Syndrome patients. This, together with a suboptimal setting of the adult care, leads to a large rate of lost to follow-up, increasing the risk for untreated comorbidities and additional costs for the health care system. On the basis of the suggestions of our adult patients, we propose a tight collaboration with a specialized endocrine gynaecologist from late adolescence complemented by an adult endocrinologist. Information about health issues and development of health care autonomy is central.

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