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 Inventor.

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 questionnaires
- 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
- 53/39 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 Gynecologist: 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

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Current medical care judged by investigators*

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Oestrogen Status objectively

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<td>39 patients</td>
<td>17 (43,5%)</td>
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Oestrogen Status subjectively

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<td>34 patients</td>
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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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