ESPE Turner Syndrome Working Group Newsletter March 2021

The purpose of this newsletter is to give active and associate members within the ESPE Turner Syndrome (TS) Working Group an update on work in progress and ongoing trials, and to give notice of the group’s participation in the forthcoming ESPE meetings.

1. Registry Platform/ Turner syndrome Registry

The Office for Rare Conditions, Glasgow held a workshop on Rare Endocrine Registries on the 12th and 13th of December 2019 in Glasgow [https://eurreca.net/rare-endocrine-registries-workshop/]. A proposal for an international Turner syndrome registry was presented.

In June 2020, an application for a grant of €15,000 from EURRECA to create a registry/platform to collect data for patients with TS has been accepted. With this registry, we aim to build an interdisciplinary, international platform that provides the means to enter standardized information to improve clinical care, to improve research activities and to understand conditions affecting Turner syndrome from a holistic point of view. The registry includes general patient characteristics (mandatory) as well as follow up data (desirable). The registry will be built as a separate module in the I-DSD registry (see also [home.i-dsd.org]).

To represent I-TS in the I-DSD platform, Prof. Dr. Claus Gravholt (endocrinologist, Denmark) will be proposed as a member of the steering committee, together with Mrs Arlene Smyth (patient representative, United Kingdom). Dr. Janielle van der Velden (pediatric endocrinologist, the Netherlands) will be part of the scientific committee, on behalf of the ESPE TS working group.

The required data to be collected have been discussed, and construction of the website on the I-DSD platform is under way. Once work on the website permits a test-site to be made available, data on some test patients will be entered so that the system can be evaluated. Information documents and informed consent forms for patients and families are being adapted from the I-DSD forms, and will need to be translated in several languages. All help for these translations will be welcome.

We aim to launch the I-TS registry in summer 2021.

2. Registry for pubertal induction oral versus transdermal 17 β-estradiol

After publication of the paper outlining pubertal induction with oral versus transdermal 17 β-estradiol in Hormone Research in 2019¹, this document, and a subsequent protocol submitted to the UK regulatory authorities² are available to be used as a basis for the management of girls requiring pubertal induction. Prospective data collection from estrogen-treated Turner patients can be entered by individual clinicians into the I-TS Turner syndrome registry once the platform has been created (see 1.).

3. Availability of 17-β estradiol (oral and transdermal)

In many countries, difficulties in obtaining 17 β-oestradiol are being reported. A particular problem concerns patients requiring doses of oral estradiol which are less than the 250 μg obtained by halving a scored 500 μg tablet (e.g. Cetura).

In the Netherlands, low-dose preparations are made up by the university hospital pharmacist, and a document on how to prepare low dose 17-β estradiol is in preparation, so that a standard procedure is available.
4. **Questionnaire concerning availability of oral and transdermal estradiol, and progestogen preparations**

In order to obtain more information of the available estrogens and prescription habits within and beyond Europe, Prof. Dr. Aneta Gawlik has created an online questionnaire for physicians. *Both ESPE and non-ESPE Members are welcome to participate in this survey.* Aneta has also prepared a questionnaire for patients and we are aiming to provide more languages for this as soon as possible.

The link to the survey for PROFESSIONALS regarding female puberty induction in Turner syndrome is: [https://www.surveymonkey.com/r/TNF3VXT](https://www.surveymonkey.com/r/TNF3VXT)

The link for Patients and Parents/Families regarding female puberty induction in Turner Syndrome is: [https://www.surveymonkey.com/r/RMNMY2G](https://www.surveymonkey.com/r/RMNMY2G)

5. **Turner Fertility Trial**

The study protocol to study the efficacy of ovarian tissue cryopreservation, discussed at the ESPE meeting 2019 in Vienna, has now been published in the British Medical Journal.

6. **Turner syndrome ‘Mini-puberty-study’: call for inclusion of patients**

For adequate counselling of patients/parents of patients with TS, early markers indicating and predicting the ovarian reserve are important. The aim of this study is to find markers of ovarian capacity, during the minipuberty, in order to predict ovarian reserve in the future. Main study parameters/endpoints: Serum levels of FSH, LH, AMH, inhibin B, testosterone and oestradiol at the age of 3 and 9 months.

This is a prospective, multi-center cohort study coordinated from the Radboudumc University Hospital, Nijmegen, The Netherlands (coordinator Dr. Janielle van der Velden), in collaboration with the University of Copenhagen, Denmark (Prof. Dr. Anders Juul). *All girls with a pre- or perinatal diagnosis TS can be included after informed consent. For inclusion, please email janielle.vandervelden@radboudumc.nl.* The first patient (Sweden) will be hopefully included soon.

7. **ESPE Turner Syndrome Working group symposium at ESPE 2021**

Due to the Coronavirus pandemic, the next ESPE meeting will again be a virtual event in September 2021. TS Working Group has been invited to organize a program lasting one hour, but it is not yet clear if this time allocation includes discussion. Details about the program follow as soon as possible.

8. **ESPE TS working group business meeting in 2021**

An online business meeting of the Turner syndrome working group is planned in September 2021. The details and agenda will be provided when available.

**References**


On behalf of the steering committee of the Turner syndrome working group, March 2021,

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