

EGS-News

No. 2

June 1999

Let's go on!!!!



Topic

Hormone
Problems
of
girls/women
with
Galactosaemia

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REPORT OF AUSTRIA

Girls with Galactosaemia very often have problems in the hormone area. The organs, in which the problems occur, are the ovaries.

The ovaries consist of, among other things, the stroma (a type connective tissue) with the follicles. In these follicles there are on the one hand hormone-producing cells and on the other hand germ cells. The germ cells are those, which are responsible for the fertility .

In the brain is the pituitary gland which produces the hormones LH and FSH (gonadotropins). These hormones stimulate the ovaries to produce the appropriate female and partially male hormones - the oestrogen, the progesterone and to a lesser degree also the testosterone. During the whole childhood the pituitary gland is there, also the cells, etc. but there is no activity.

Normally the pituitary gland starts very strong production of LH and FSH in puberty and the ovaries are stimulated to produce hormones. Above all oestrogen causes these modifications, which one calls puberty development, i.e. the formation of the bosom, and the build up of mucous membrane in the uterus which is rejected cyclically. Usually the first signs of puberty development are noticed at 11 - 13 years of age. The bosom grows, about 1 1/2 – 2 years later the first menstruation begins. If the ovary functions normally, not only is the oestrogen formed, but also the progesterone. Consequently, approx. 1 - 2 years after the first menstruation a regular production of oestrogen and progesterone connected with an ovulation begins . Starting from this point in time a girl is fertile.

The oestrogen is also responsible for the correct bone density .

When girls have Galactosaemia, the hormones LH and FSH do work, but the ovaries do not react. The hormones are blocked and the hormone-values therefore increase. That is the reason why there is no production of oestrogen in the ovaries, that the growth of the uterus is missing, that the menstruation is missing and also the clinical puberty development. The pubic hair will be normal, because there are other hormones responsible for this.

The few studies, which look into the subject of hormone problems with Galactosaemia, report that 75 - 90 % of all girls and women with Galactosaemia have a problem with the ovaries.

According to the research with rats it is likely that the toxic effect on the ovaries already occurs in the foetus before the birth . Even a lactose-free diet of the mother during the pregnancy could not prevent the problems.

The puberty development usually begins at the age of 11 years, but it can also be at the age of 8 or 14 years. If a 13 - 14 year old galactosaemic girl has no puberty development you should check the hormone situation. This missing puberty development probably affects her size. Children, who do not enter puberty, become also temporarily, but by therapy influenceable, smaller than girls in the same age, who already are in puberty. Therefore it is meaningful to document the size and to make also a bone age examina-

tion with the help of a hand root X-ray.

Which therapy possibilities are there?

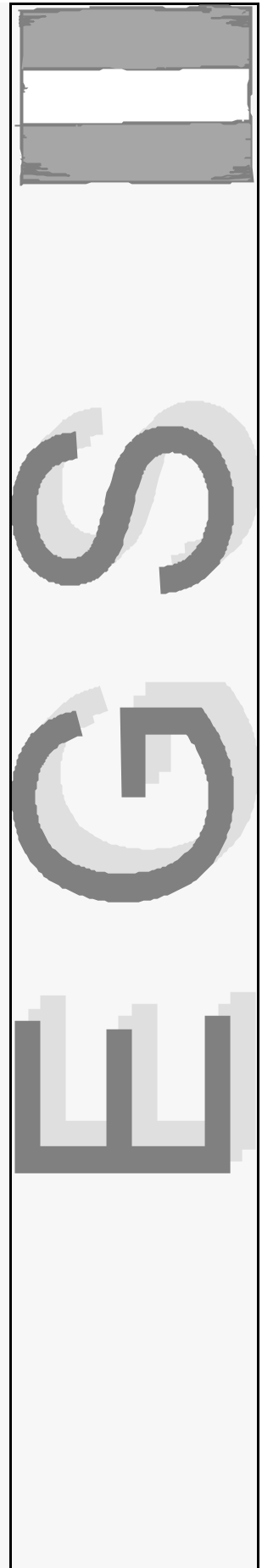
When the puberty development is absent (approx. at 14 years old), the values of oestrogen/gestagen are abnormal, menstruation is increasingly irregular and the bone density is reduced as if there is an early Menopause, you should start a hormone replacement. First you will begin with pure oestrogen in small doses. After 1 - 1 1/2 years and after the first menstruation the oestrogen will be combined with gestagen, so that correct cycles occur.

Which hormone preparations are there?

- tablets, which possibly contain lactose in small quantities.
- Plaster patches/pads - here the hormone penetrates through the skin into the body and
- for gestagen also pessaries

The oestrogen is also important for well-being (sexuality) and bone density.

The article is an abstract and a translation of the speech of Doz. Dr. Gabriele Häusler, AKH, Vienna, held at the general meeting of the ÖGAST in May 1997



REPORT OF FRANCE

My daughter Marie was born the 26th of February 1985. She was a tiny, slender girl up to the age of 11. She was breastfed at the beginning. Her growth was sometimes painful, and Marie used to complain about it.

The last two years, her body changed a lot. She grew taller, she lost the figure of a little girl to take the figure of a woman; her waist is more obvious, so are her hips; nobody can fail to notice her breasts now. Her pubic and auxiliary hair is present too.

She had her first period in February 98 when she was exactly 13. Until now, it is almost always regular. Marie also complains about feeling a pain before the periods, and when she has them. The doctor who follows her for Galactosaemia confirms she is growing up normally like a non-galactosaemic adolescent.

Catherine Moniez

REPORT OF GERMANY

Lieselore Rohlfing/Germany asked Doz. Dr. Susanne Schweitzer to comment on the subject:

It is well known that about 80% of the girls suffer from a subnormal function of the ovaries due to fibrosis which is perhaps already caused in the womb. To start puberty the brain releases follicle stimulating hormones (FSH) in order to produce estradiol (EL) in the ovaries.

Since this doesn't happen (Hypogonadism) even more FSH is released (Hypergonadotrophy).

And if there is no oestrogen production there will be no development of the breast - only the growth of pubic hair. In addition to that the ovaries won't develop - hence no menstruation. In subsequent years a osteopenia (reduction of bone mineralisation) is caused.

Puberty can be started if oestrogen is given orally or per transcutaneously (through the skin-patches). Sometimes an oestrogen-gestagen combination is preferred. If this hormonal substitution is stopped there will be a bleeding due to the omission.

The fact that these girls are less fertile can't be changed by providing these hormones. The egg donation of a fertile woman - in-vitro-fertilisation with the sperm of the galactosaemic woman's partner and the implantation of a fertilised ovum into the uterus of the GAL woman can be carried out e.g. in the USA.

It is very important that the children - and adult endocrinologist and the gynaecologist work in collaboration.

REPORT OF GREAT BRITAIN

Caroline Duker by Steve and Terry Duker

Our daughter Caroline was born in November 1982 and diagnosed with galactosaemia on day 7. She had been transferred from our local hospital to Great Ormond Street Children's Hospital in London, UK for diagnosis and treatment and was being cared for by Professor James Leonard.

From the very start, Professor Leonard explained the possible problems we might experience with regard to Caroline's educational achievements, possible infertility and hormone treatment.

Caroline had a pelvic scan and blood tests to ascertain whether or not her ovaries were working at 9 years of age. The results showed that she had some ovarian tissue but that the ovaries were not working properly. These tests were repeated at 11 when they told us that there were no follicles to produce eggs. This would mean that if Caroline decided to have a baby later in her life, it would have to be by the means of a donated egg. It was decided that now was the time to start hormone replacement treatment.

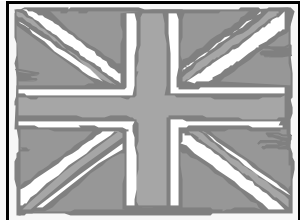
She started on a very low dose of ethinyloestradiol (2 micrograms) and gradually increased the dose over the next 2 years. However, after 2 years she had not developed as much as was expected and the decision was made to go back to the beginning and start on 5 micrograms. This time it was agreed to leave Caroline on each dose for a longer period of time to enable maximum breast growth.

Caroline's endocrinologist is very happy with her growth and is confident that once she begins to gain weight, this will maximise her breast development.

Caroline is currently taking 10 micrograms of ethinyloestradiol every day and for 10 days a month she takes norethisterone, which ensures that she has a monthly cycle, otherwise she would bleed continuously. We expect that next time we visit the hospital in May 1999 for her six monthly appointment, the endocrinologist will change Caroline's hormone treatment to the next stage.

Although the tablets Caroline takes every day contain lactose, there are no hormone tablets available that do not contain lactose. Hormone patches do not contain lactose but they are only suitable for adults who are on full dose hormone replacement. We were assured that the amount of lactose in these tablets is insignificant when taking into account the amount of galactose the body makes itself.

Caroline is happy taking the tablets and is coping well with the monthly bleeding which only lasts 1-2 days.



REPORT OF THE NETHERLANDS

Hello, I am a 32 year old woman with Galactosaemia!

Ten days after I was born in 1967, I was hospitalised for what later turned out to be a rare disease called galactosaemia. Except for the fact that I had to keep my diet, I grew up just like other kids, without any major problems caused by galactosaemia. So I hadn't been seeing doctors for regular check-ups since I was about 14 years old. And I was unaware of any hormonal problems that might occur later on.

I had my periods, although they were extremely irregular. Sometimes two months passed, sometimes even as much as six months. At the time neither myself nor the doctor combined these problems with galactosaemia. The doctor prescribed the pill, and I got regular bleedings because of this.

A couple of months after I got married at the age of 27, my brother and his wife were expecting their first baby. They wanted to know if there was a chance that their baby might have galactosaemia. New tests were run and this is when I found out about hormonal problems in relation to galactosaemia. My uterus turned out to be very tiny, but that wouldn't necessarily cause further problems. What was worrying was the fact that hormonal activity was as low as in women that go through menopause and therefore I was told I could not have children of my own.

After a few months we applied to adopt a baby from a foreign country. First there was a waiting period of over a year. Then there were 6 biweekly sessions held by the Dutch Ministry of Justice to prepare people for adoption. Finally we had a couple of sessions with a social worker and we obtained the official approval for adoption. The next step to be taken would be to find an adoption organisation and make the final arrangements.

We never took that last step. I'm not sure if it was due to personal problems or some other factor. Right now we think that maybe having kids is not the best thing for us.

Recently a little miracle happened. I didn't feel like taking the pill, and after about 3 weeks without it, I got my period. And after that again. Now I'm in the middle of new medical tests. Blood tests have shown that there really is some hormonal activity, but it is still at a much lower level than in non-galactosaemic women of my age. It's too early yet to even really think about possible consequences. For now I'm just happy that my body is doing this on it's own. And only time will tell what happens next.

March 1999, The Netherlands

REPORT OF THE SWITZERLAND

Hormone-Treatment of young women in Switzerland

As there are only a few young ladies with Galactosaemia in Switzerland, the results of our enquiries are not representative. Hereafter the replies to our questions:

Start of the therapy? - at the age of 16½ (A) and 18 years (B)

Reasons to start therapy?
- Ovarian failure due to Galactosaemia.
- Retarded development of the body
- No menstrual period

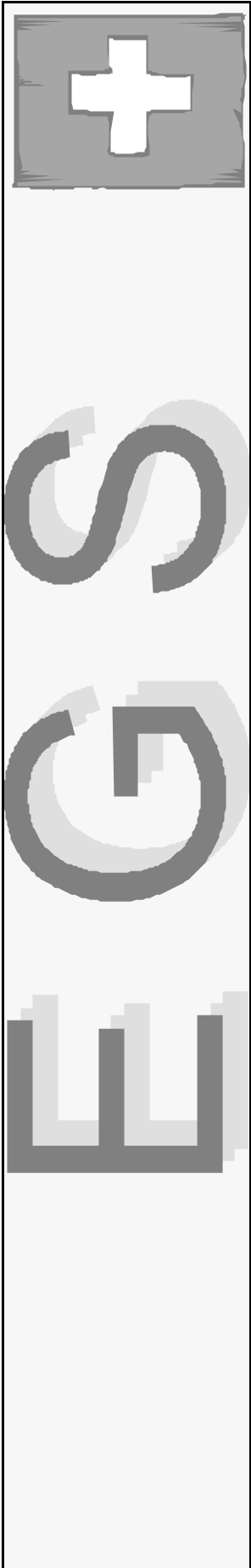
Medical treatment **A:**
for 3 months hormone Pads with Estraderm TTS 25, first every second day, after 1 month daily, since then: Estracomb TTS (transtermales therapeutic system with Oestradiol, first 4 mg, afterwards 10 mg and Noretisteron-Acetete 30mg)

B:
Hormone pads Estraderm MX 50
As all the necessary hormones are not included an additional injection of hormones has to be given every three months

Effects: **A:**
Thanks to the hormone pads, **A** had a harmonious puberty, she grew and put on weight (170 cm/47 kg). She has a normal menstrual period. She developed into a young woman. At the age of 19, the duration of **A**'s menstrual period increased to ten days. Her body was under stress. She felt tired and weak. Therefore she now gets an additional medicine (Prodafem 5 mg) a few days before her period. Unfortunately, these pills contain a small amount of galactose. This is an experiment of a limited time.

B:
No secondary effects.

March 1998, C. Gutknecht



Who's Who at the EGS?

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Secretary Maaïke van Kempen, Netherlands
Treasurer Steve Duker, Great Britain

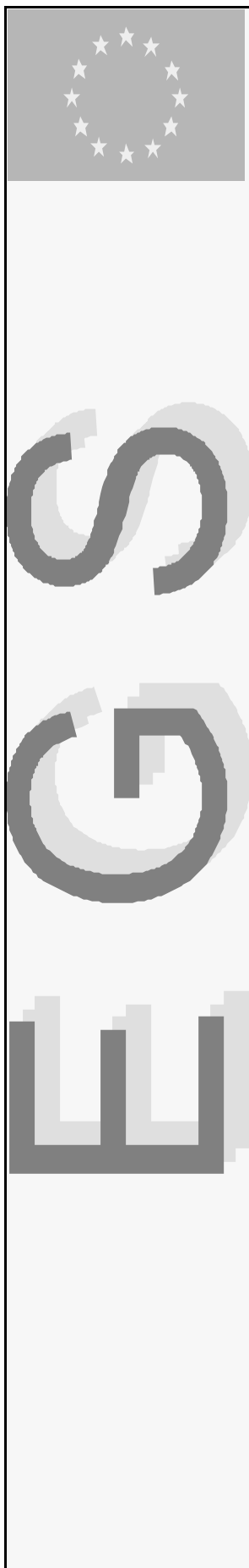
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