

Galactosemia Foundation Conference 2012

Reproductive System Update

Issues for females

Premature Ovarian Insufficiency

Research in the US has shown that Premature Ovarian Insufficiency (POI) is the only symptom of galactosaemia that is experienced by almost all patients. Some researchers mentioned that they have never come across a female with galactosaemia that has not experienced POI, while others said that the rate was certainly greater than 80% and probably up to 95-99%. POI is defined as menopause in a woman up to 40 years old.

For some women and girls the problem is described as Primary POI, which means that external hormones are needed to induce puberty. The delivery of oestrogen to these patients is likely to need to be continued throughout the period of life before which the general population would experience menopause (i.e. until the late 40s or early 50s). This is because oestrogen is important for bone strength and heart function. The time and method of inducing puberty in girls experiencing Primary POI will depend on a number of factors and parents and girls concerned with this should discuss it in more detail with their healthcare providers. The general consensus from presenters at the conference was that if a girl is not showing any signs of puberty by 12-13 years old serious consideration should be given to inducing puberty. However, if some signs of puberty are present at that age, it would be ok to wait until the girl is 15 years old.

For others the problem is Secondary POI, which means that puberty is achieved naturally, but that at a point in time before 40 the menstrual cycle stops. Hormone therapy for women in this group would need to be discussed once it is recognised that the cycle has stopped.

Researchers have been working to determine why this difference exists. At present it appears that there may be a link between residual GALT activity and the health (or otherwise) of the ovaries. As you may be aware galactosaemia is caused by different gene mutations that are passed on from both parents to the child with galactosaemia. Some of these gene mutations result in zero GALT activity, while others leave some residual GALT activity. Some studies show that there appears to be a correlation between residual GALT activity and AMH levels. AMH is a hormone typically seen in high levels in girls and pre-menopausal women). Women with galactosaemia who have relatively high levels of AMH

(when compared to other women with galactosaemia) are more likely to reach puberty naturally. However, much more work needs to be done and there is certainly no guarantee that residual GALT activity will result in natural puberty or healthier ovaries.

Pregnancy

Rare cases have been seen throughout the world where women with galactosaemia have been able to conceive and carry babies to term. However, for the vast majority of women with galactosaemia this is a very unlikely outcome.

Research has shown that in most females with galactosaemia the ovaries suffer significant damage either before or shortly after birth. This means that some females will never produce a healthy egg. However, a small number of females with galactosaemia will produce some eggs, although even in these patients the number of eggs is likely to be significantly less than seen in the general population.

The possibility of egg harvesting from girls with galactosaemia for freezing and use later in life was discussed. This is a controversial topic and opinion was divided amongst attendees. It is particularly controversial for galactosaemia because there is no guarantee viable eggs will be recovered given that the damage can happen very early in life (well before egg harvesting could be contemplated) and, in any event, current IVF techniques have shown limited success in this respect.

Alternatives to egg harvesting were discussed and it was clearly stated that there is no reason why a woman with galactosaemia cannot carry a baby conceived via IVF using a donor egg from another woman. The other obvious alternative is adoption which, although seemingly more common in the US than Australia, is something that is able to be presented to girls with galactosaemia as a path to them one day having a family of their own despite the obvious challenges.

A helpful guide for parents and women dealing with galactosaemia and POI that was mentioned at the conference can be found at:

http://newenglandconsortium.org/brochures/Your_Guide_to_Galactosemia_and_POI.pdf

Issues for males

Traditionally the focus of fertility issues with respect to galactosaemia has been on the effect of galactosaemia on the female reproductive system. However, researchers have now given some consideration as to whether there is also an impact on males.

The good news is that the male reproductive system is not affected to anywhere near the same degree as the female reproductive system. However, research has shown that sperm count in males with galactosaemia tends to be in the low

normal range, semen volume can be reduced and that in adult males studied there are fewer instances of males becoming biological fathers than in the general population. The reasons for this are currently not fully understood and further research needs to be done before any conclusions can be made about the impact that galactosaemia has on male reproduction.