

## Premature ovarian insufficiency:

### why is it not being diagnosed enough in primary care?

#### WHAT IS IT?

How good are we in primary care at diagnosing and managing premature ovarian insufficiency (POI)? Unfortunately, not very, as this condition is still underdiagnosed and undertreated. Many GPs have not heard of this condition and admit to not being confident diagnosing it. POI is very common; it affects 1% of women under 40 years and 0.1% of women under 30 years in the UK.<sup>1</sup> Interestingly, 2.5% of all patients with POI are adolescents. One reason for an increasing incidence in this condition is that there is increased survival from childhood cancers and many of these women have received radiotherapy and chemotherapy, often resulting in adverse effects on their ovarian function.

POI is defined as a loss of ovarian activity before the age of 40 and is characterised by irregular or absent periods and reduced fertility. Symptoms of oestrogen deficiency often occur, which include hot flushes, night sweats, mood changes, memory problems, vaginal dryness, dyspareunia, and low libido.<sup>2</sup> It is most commonly idiopathic.

We have both seen many women who have been told by doctors that they are *'too young to be menopausal'*, or that *'missing periods is nothing to worry about and certainly does not need any treatment'*.

#### WHY IT'S SO IMPORTANT TO DIAGNOSE POI

So why is it so important that these women are diagnosed promptly? Clearly it is important that their symptoms are addressed as these will often improve dramatically with the right type and dose of hormones.

However, the most important reason for diagnosing these women promptly is that both mortality and morbidity in women with untreated POI is increased. The reduced life expectancy is largely due to cardiovascular disease.<sup>3</sup> There is also an increased future risk of osteoporosis in these women if they are not given treatment. A timely diagnosis and initiation of treatment will reduce the impact of future complications from POI.<sup>4</sup>

A history of menstrual disturbance

(especially oligomenorrhoea or amenorrhoea) in woman under 40 should alert healthcare professionals to consider prompt testing for POI by measuring serum FSH levels. More than one FSH level should be undertaken for the diagnosis to be made.<sup>5</sup>

Infertility is a major complication of POI, so referral to a fertility centre may be indicated and the best results for a live pregnancy are often as a result of egg donation.

Treatment with hormone replacement therapy (HRT) should be started and continued until at least the average age of menopause (51 years). Hormone replacement is important for treating symptoms and protecting against the long-term effects of oestrogen deficiency including cardiovascular disease, osteoporosis, and cognitive impairment.<sup>1,5</sup>

Women should be reassured that there is no increased risk of breast cancer in taking HRT at a young age as they are simply replacing hormones that their body should otherwise be producing.

A frank discussion needs to take place about whether the patient wants to continue with HRT after she reaches the age of the 'normal' menopause. The length of time on HRT in this population of patients again is not relevant and only starts becoming significant once they reach the age of 51.

Some women with POI who need contraception do take the combined oral contraceptive pill (COC); this will provide hormone replacement and contraception. Women are advised to take it continuously or with long cycles of at least 3 months to minimise the periods of oestrogen deprivation.<sup>5</sup> However, the beneficial effects of taking the COC on bone mass density and lowering cardiovascular risk are less favourable than with taking HRT.<sup>6</sup>

Sexual dysfunction is very common in women with POI and needs to be asked about directly. Local oestrogen treatment may be very helpful for vaginal dryness and urinary symptoms and can be taken in addition to systemic HRT. Low libido is common and

testosterone is often beneficial, although it is not currently licensed for women in the UK. Treatment with testosterone should be evaluated after 3–6 months and should possibly be limited to 24 months in the absence of long-term safety data. Women need to be counselled about this so that they can make an informed choice.<sup>5</sup>

The diagnosis of POI for a young woman can be devastating and supportive counselling can be very important and helpful.

Making the diagnosis of POI early and offering appropriate treatment can therefore often reduce or prevent long-term sequelae from POI. Women should be reassured about the importance of hormone treatment.

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