

# Are expanding disease definitions unnecessarily labelling women with polycystic ovary syndrome?

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## Re: Are expanding disease definitions unnecessarily labelling women with polycystic ovary syndrome?

Copp et al contended that polycystic ovary syndrome (PCOS) is over-diagnosed, recommending a delayed approach to diagnosis and potential treatment of symptoms without a diagnosis (1). This was proposed largely based on uncertainty of diagnosis in adolescents and expanded PCOS diagnostic criteria with multiple phenotypes, some of which may have more limited long-term health effects. Challenges in PCOS diagnosis in adolescents are acknowledged and over-diagnosis is an important consideration in any field, however here a fundamental principle of clinical care was ignored – the experience of the patient. The BMJ commentary failed to engage affected women or to consider the significant evidence regarding diagnosis experience from studies involving women with PCOS.

In a commentary in the Medical Journal of Australia (2) patient advocates, healthcare providers and academics contested the position put forward by Copp et al. Here we summarise this evidence-based counter-case, showing that PCOS is under-recognised and that delays in diagnosis and inadequate information provision are distressing and unacceptable to women with PCOS. We also describe an international initiative aiming to address challenges in PCOS constructively and through partnership with patients, to improve PCOS diagnosis, patient experience, education and health outcomes.

Copp et al suggest that adoption of the internationally endorsed Rotterdam diagnostic criteria has contributed to an increase in PCOS prevalence from 4-6% to 21% (1). We argue that these are selected and extreme estimates drawn from disparate populations; 21% using the Rotterdam criteria comes from a study reporting prevalence of 15% using the original National Institutes of Health (NIH) criteria, in a population with a high risk of metabolic conditions (3). Instead, systematic review evidence shows more a limited impact of diagnostic criteria on prevalence (NIH criteria: 6% and Rotterdam criteria: 10%) (4). It is important to continue to research the ideal cut-offs for each PCOS diagnostic criterion, but we contest that selective reporting of prevalence does not provide evidence of over-diagnosis.

There is also no direct evidence from studies of clinical care that PCOS is over-diagnosed. Rather, evidence from hospital and primary care settings indicate that many cases of PCOS go undetected (5, 6). Community-based studies also suggest PCOS is significantly under-recognised (7).

Copp et al argue that a diagnosis of PCOS may cause women with non-hyperandrogenic phenotypes needless distress, due to potentially incorrect perceptions of metabolic risk (1). While further research is required regarding the long-term health outcomes associated with the different PCOS phenotypes, insulin resistance occurs in 75% and 95% of lean and overweight young women with PCOS (8), with current evidence suggesting limited impact

of diagnostic criteria (9). Furthermore, differences in cardiometabolic risk observed across PCOS phenotypes may be largely attributable to differences in adiposity (10). We fully support that more research is needed; however, in the interim, we recommend that informing women about potential variation in metabolic features across the phenotypes is preferable to delaying PCOS diagnosis.

It is internationally accepted that a cautious approach to PCOS diagnosis is recommended for adolescents (11). However, the proposal to delay PCOS diagnosis more broadly (1) neglects the consistently reported experiences of women with PCOS. Mrs Veryan McAllister, President of the Polycystic Ovary Syndrome Association of Australia relates her own experience: "The assertions by Copp et al are extremely upsetting to read from a patient and patient advocacy perspective. As a patient I had to see multiple health care providers, my own diagnosis was delayed six years and caused extreme anxiety, and I am not alone. The labelling of the disease helped me to realise it was real and that there were steps I could take to control it. Whilst this is only my story, it is supported by evidence on women's experiences from around the world".

Internationally, women with PCOS report commonly seeing numerous healthcare providers about PCOS symptoms, raising their concerns repeatedly, and experiencing difficult delays before receiving a diagnosis (12-17). Longitudinal data suggests that women with PCOS had equally high levels of distress in both the 12 months before and after PCOS diagnosis (18), suggesting distress was related to the features of PCOS rather than receiving a diagnosis. We recognise that a diagnosis can be confusing, worrying or lead to women feeling a lack of control, if support and information are not provided (14-16). Health care providers are the initial and trusted source of information (16, 17), yet evidence shows we are already letting women down in this regard (12, 13). It is difficult to see how delaying diagnosis and treating isolated features of PCOS without a diagnosis or opportunity for education, will address these gaps. Instead, diagnosis creates an important opportunity to provide personally relevant information and enable women to feel more in control of the condition.

The first international evidence-based guideline for PCOS will be launched in 2018 (<http://www.pcos-cre.edu.au/evidence-based-guidelines-polycystic-ovary-sy...>). This unique initiative is built on input from thousands of affected women and health care providers, aims to address challenges in diagnosis and inconsistency in PCOS care, and will be accompanied by extensive implementation and evaluation programmes. The initiative is led by the NHMRC Australian Centre for Research Excellence in PCOS, in partnership with the European Society of Human Reproduction and Embryology, and the American Society for Reproductive Medicine, with over 30 other societies and patient advocacy groups engaged. We believe this will provide a more constructive, evidence-based and co-developed approach to improving PCOS diagnosis and healthcare and propose that suggestions to delay PCOS diagnosis in adult women are unfounded and likely to only increase confusion and distress.

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