# Letters

## Reproductive carrier screening

The July 2018 issue of *Australian Journal of General Practice* included an article on preconception care.[1] The intent of the article was laudable, but I am concerned that the recommendations regarding genetic carrier screening do not reflect current advice in this field.

A family history of heritable disease is an important factor that should be addressed for a couple. However, the absence of such a history does not provide much reassurance. The family history of a child with a recessive disorder (either X-linked or autosomal) is typically unremarkable.

Recessive disorders represent a significant consideration in reproductive planning. The chance of having a child with cystic fibrosis, spinal muscular atrophy or fragile X syndrome is similar to the population chance of having a child with Down syndrome.[2]

Reproductive carrier screening is increasingly recognised as an effective way of reducing that possibility. It is not necessary that all such testing be managed through specialised genetic services; GPs have been managing carrier screening for thalassaemia (another recessive disorder) for years.

Genetic carrier screening for these common recessive disorders is provided by a number of pathology laboratories in Australia. Many laboratories also provide haematological studies to identify carriers of thalassaemia. These laboratories provide genetic tests in the context of a medical consultation, not as 'direct-to-consumer' tests.

The latest Royal Australian and New Zealand College of Obstetricians and Gynaecologists/Human Genetics Society of Australasia guidelines on carrier screening note that 'information on carrier screening for the more common genetic conditions that affect children (eg cystic fibrosis, spinal muscular atrophy, fragile X syndrome) should be offered to all women planning a pregnancy or in the first trimester of pregnancy'.[3] The Royal Australian College of General Practitioners guidelines concur, noting that doctors should 'provide opportunity for carrier screening for genetic conditions (eg cystic fibrosis, haemoglobinopathies)'.[4]

Offering reproductive carrier screening prior to conception simply represents good medical practice.[5]

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### References

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2. Archibald AD, Smith MJ, Burgess T, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: Outcomes of 12,000 tests. Genet Med 2018;20(5):513–23. doi: 10.1038/gim.2017.134.
3. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists, Human Genetics Society of Australasia. Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions. East Melbourne: RANZCOG, 2018.
4. The Royal Australian College of General Practitioners. Guidelines for preventive activities in general practice. 9th edn. East Melbourne: RACGP, 2016.
5. The Royal Australian College of General Practitioners. Genomics in general practice. East Melbourne: RACGP, 2018.

## Reply

Thank you for your letter regarding genetic screening for recessive conditions in preconception care. The past decade has seen an impressive evolution in genetic technology, with multi-panel screening tests available for common recessive conditions that affect one in 20 Australians.[1] This has been accompanied by increased public awareness in the wake of a call to make this testing free by the parents of a child born with spinal muscular atrophy (SMA). The Australian Government has recently announced they will fund a trial, the Australian Reproductive Carrier Screening Project (ARCSP), that will include screening for SMA, cystic fibrosis and fragile X syndrome.[2] During the project, '10,000 Australian couples will be screened before they conceive or in early pregnancy. These couples will be offered information and support to help them make informed choices about whether or not to have the test', and affected couples identified in the preconception period would be offered subsidised in-vitro fertilisation treatment.

This is a terrific initiative; however, at present, screening for low-risk couples is at the couples' own expense. Currently, individuals are required to pay approximately $400 for screening for these three recessive conditions. Additionally, outside of the ARCSP trial, preimplantation genetic diagnosis is currently not covered by the Medicare Benefits Scheme. The Royal Australian College of General Practitioners' preventative care guidelines, which we consulted, state that general practitioners should 'provide opportunity for carrier screening for genetic conditions (eg cystic

fibrosis, haemoglobinopathies) and referral for genetic counselling based upon risk factors'.[3] The Royal Australian and New Zealand College of Obstetricians and Gynaecologists acknowledges that carrier screening is better conducted in the preconception period, but also acknowledges that this is not publicly available for low-risk women.[4] We agree that it is reasonable to provide information about screening to women or couples without a family history of genetic conditions, but that they should be informed of the benefits, limitations and cost of screening.

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## No more *P* values

The *Australian Journal of General Practice* has recently published an article by Tam et al, 'How doctors conceptualise *P* values: A mixed methods study' (*AJGP* October 2018).[1] The authors explain in this article, and in associated online materials (available at https://vitualis.com/?p=2625 and www.youtube.com/watch?v=kK6I1KCb7jQ), the inappropriateness of quoting *P* values in reports of studies.

This leads me to ask whether the journal will now instruct authors to give confidence intervals for their findings and not to quote any *P* values. When submitted manuscripts contain *P* values, will the editors ask the authors to remove them and replace them with confidence intervals?

The journal would do well to remind authors and readers that effect size is still the most important element in the results, that it needs to be large enough to be of clinical significance, and that when two results are clearly different, no statistical testing is necessary to demonstrate or confirm that difference.

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**Reference**

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