

Case 26 – Mark

Mark's father is suffering from Huntington's disease. Mark has decided that, in spite of the fact that he knows he has a 50% chance of being a carrier of the gene causing the disease, he prefers to live with the uncertainty. In other words, he does not want to have a test to find out if he carries the Huntington gene. (Around 80 to 90% of the population who are in a similar position share Mark's preference). His wife, Kate, however, feels that she would like to know whether or not Mark is a carrier as they are contemplating starting a family.

Best Practice Response

- Mark and Kate need to be thoroughly informed and well supported. While a GP can discuss the situation with Mark and Kate, it would be in their best interests for the GP to refer them to:
 - A genetic counselling service (usually including a counsellor and a geneticist) to help them work through the issues for both of them
 - A support organisation (there are Huntington's Disease organisations for each state and territory)
- The long-term consequences of a positive test can be devastating for patients and their families. There may be severe psychological consequences such as depression, anxiety, survival guilt, and suicide (1), as well as potential discrimination in insurance or employment. Studies of people deciding whether to have the test have found a key factor is doing the right thing for their children – but this can direct them either towards or away from testing (2).
- Even if Mark had the test he may decide not to disclose the results to his wife.
- In Australia doctors are legally able to disclose genetic information to relatives, subject to strict regulation as specified in NHMRC guidelines (3) and Australian Privacy Principle 6.51 and 6.53 (4). The doctor must believe that the disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative. Note that the recipient of the information must be a genetic relative of the individual, and Kate is not a genetic relative of Mark.
- In May 2017 the UK Court of Appeal announced that a trial will go ahead in which the daughter of a man with Huntington's Disease is suing the hospital caring for her father, claiming that the hospital had a duty of care to share the father's Huntington's Disease diagnosis with her, against her father's wishes, so that she could seek out her own genetic testing (and potentially have a termination of pregnancy).

- (1) Coustasse A, Pekar A, Sikula A, Lurie SJ. *Ethical considerations of genetic presymptomatic testing for Huntington's disease*. Hosp Mark Public Relations. 2009 Jul-Dec;19(2):129-41. PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19827323>
- (2) Smith JA, Stephenson M, Jacobs C, Quarrell O. *Doing the right thing for one's children: deciding whether to take the genetic test for Huntington's disease as a moral dilemma*. Clin Genet. 2013 May;83(5):417-21 Medline: <https://www.ncbi.nlm.nih.gov/pubmed/23438681>
- (3) National Health and Medical Research Council. *Use and disclosure of genetic information to a patient's genetic relatives under Section 95AA of the Privacy Act 1988 (Cth) – Guidelines for health practitioners in the private sector*. 2009; NHMRC: Canberra. Pdf available at https://www.nhmrc.gov.au/files_nhmrc/file/publications/guidelines_under_s95aa_use_of_genetic_information_2014.pdf
- (4) Office of the Australian Information Commissioner. *APP Guidelines: Chapter 6: APP 6 – Use or disclosure of personal information*. 2014. Pdf available at <https://www.oaic.gov.au/agencies-and-organisations/app-guidelines/chapter-6-app-6-use-or-disclosure-of-personal-information>

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