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Genetics in the Adult: Assessing Risk

Practice Based Small Group Learning Programme

INTRODUCTION

Requests from patients for assessment of genetic risks and subsequent testing are on the rise. So too is interest in direct-to-consumer testing. Applying genetic concepts in practice, however, is complex. Assessment of risk, benefits and limitations of testing, interpretation of test results, and potential implications for insurance and employment are some of the challenges involved.

OBJECTIVES

This module will enable clinicians to:

- Take and interpret a family history that allows determination of individual and family genetic risk.
- Identify patients at increased risk of genetic cancers who may benefit from formal genetic counselling and discuss the implications of test results.
- Effectively counsel patients regarding direct-to-consumer genetic testing.
- Direct patients to reliable resources related to genetic risk and testing.

Notes:

- This module will focus on the non-pregnant adult and excludes paediatric and prenatal genetic screening.
- All conditions that might be candidates for genetic testing cannot be covered in their entirety here. This module includes a sampling of issues relevant to primary care that emphasize several general principles as well as the specific conditions highlighted in our cases – inherited breast/ovarian cancer and inherited thrombophilia (factor V Leiden).
- There is wide variation in genetic resources across the country. It is important to be aware of the resources available in your area.

CASES

Case 1: Natalie, age 39

Natalie is new to your practice and you have only seen her once or twice for minor illnesses. She is not on any medications, is a non-smoker and has a BMI of 28. She is married and had a tubal ligation five years ago. She has two children and works as an accountant.

Today she comes to see you because she had some severe breast pain a few weeks ago, which has since resolved, but she is concerned because there is “a lot of cancer” in her family.

What further information would be useful?

Part Two

Natalie describes herself as Caucasian and Scottish. She has a maternal aunt who had a diagnosis of breast cancer at the age of 68. She is currently doing well five years post diagnosis, although she is estranged from the rest of the family.