MSD AstraZeneca

CECOG
Central European Cooperative Oncology Group

A Guide To Genetic Consenting

Module 2:
Why is genetic consenting important?

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Learning objectives

The primary purpose of this learning module is to inform and educate non-genetics HCPs on genetic consenting.

This module will enable HCPs to:

- Be aware of the familial implications of a genetic test
- Understand how genetic testing can guide treatment decisions in cancer

HCP=healthcare professional

Genetic test results have a familial implication

For example, BRCAm results would indicate predisposition to further cancers

However, when thinking about inherited mutations, a positive BRCAm result will have an impact on other biological relatives

- For instance, a 17-year-old woman may have inherited a mutation and be at high risk of breast and ovarian cancer
- A father may have the mutation, and be at increased risk of male breast cancer and prostate cancer
- The siblings of both parents are potentially at risk of having the mutation

These aspects should be discussed with any patient undergoing testing, who should also be offered support to discuss the potential risks with others in the family

BRCAm=*BRCA1/2* mutation

Speaker communication

Genetic testing can inform treatment options in addition to identifying at-risk family members

- Results from genetic testing can inform treatment decisions, involving:

Surgery

Pre-operative testing can impact surgical decision-making in patients with gBRCAm breast cancer with the majority opting for bilateral mastectomy once they are aware of their gBRCAm status.[1]

Chemotherapy and targeted therapy

Patients with BRCAm have been shown to be sensitive to platinum across a range of cancers, including ovarian, pancreatic and metastatic breast cancer.[2-5]
Patients with BRCAm have also been shown to be sensitive to PARP inhibitors. [6-12]

- Furthermore, germline testing of cancer patients helps:

Identify at-risk family members

Identify potential future cancers in patients

This can influence the implementation of risk-reducing and prevention strategies, and screening and monitoring measures, such as:[13]

- Risk-reducing mastectomy and bilateral salpingo-oophorectomy for individuals with a known gBRCAm, whether or not they have a personal history of breast cancer[14]
- Annual radiological surveillance and chemoprevention for individuals who choose not to have a bilateral risk-reducing mastectomy[14]

BRCAm=*BRCA1/2* mutation; gBRCAm=germline *BRCA1* and/or *BRCA2* mutation; HER2=human epidermal growth factor receptor 2; HRD=homologous recombination deficiency; HRRm=homologous recombination repair gene mutation; PARP=poly(adenosine diphosphate ribose) polymerase

1. Yadav S, et al. *Hered Cancer Clin Pract.* 2017;15:11; 2.Pujade-Lauraine E, et al. *Lancet Oncol.* 2017;18:1274–1284; 3. Coleman RL. *Lancet.* 2017;390:1949–1961; 4. Golan T, et al. *Br J Cancer.* 2014;111:1132–1138; 5. Collet L, et al. *Cancers.* 2022;14(3):599 6. Paluch-Shimon S. *Breast.* 2017;35:203–217; 7. Tan DSP, Kaye SB. *Am Soc Clin Oncol Educ Book.* 2015:114–121; 8. Robson M. *N Engl J Med.* 2017;377:523–533; 9. Litton J. *N Engl J Med.* 2018;379:753–763; 10. de Bono J, et al. *N Engl J Med.* 2020;382:2091–2102; 11. Golan T, et al. *N Engl J Med.* 2019;381:317–327; 12. Rose M, et al. *Front Cell Dev Biol.* 2020;8:564601; 13. Hallowell N. *Fam Cancer.* 2019;18:293–301; 14. UK National Institute for Health and Care Excellence. Familial breast cancer: Classification, care and managing breast cancer and related risks in people with a family history of breast cancer. Clinical guideline [CG164]. Available at: https://www.nice.org.uk/guidance/cg164 (Accessed June 2022)

Summary

- Genetic test results (e.g. a pathogenic *BRCA1/2* mutation) may have familial implications[1]

- Genetic testing can inform treatment options, including surgery as well as choice of medical therapy[2,3-5]

1. UK National Institute for Health and Care Excellence. Familial breast cancer: Classification, care and managing breast cancer and related risks in people with a family history of breast cancer. Clinical guideline [CG164]. Available at: https://www.nice.org.uk/guidance/cg164 (Accessed June 2022); 2. Yadav S, et al. *Hered Cancer Clin Pract.* 2017;15:11; 3. Pujade-Lauraine E, et al. *Lancet Oncol.* 2017;18:1274–1284; 4. Coleman RL. *Lancet,* 2017;390:1949–1961; 5. Golan T, et al. *Br J Cancer.* 2014;111:1132–1138.