



Medicare item numbers for cancer gene testing: Information for clinicians

From November 1, 2017 new Medicare item numbers are available for some cancer gene tests and can be ordered directly by **specialists or consultant physicians** without a referral to Genetic Services of WA (GSWA).

What has not changed?

- Criteria for cancer gene testing under the new item numbers have not changed, and are consistent with current national guidelines (www.eviQ.org.au).
- GSWA continues to accept referrals for genetic counselling and provide genetic testing to people who meet these guidelines – these services are free to patients who have a valid Medicare card.
- Genetic testing remains available to anyone through private providers, and we recommend using a service that includes genetic counselling.
- Genetic counselling is particularly recommended for people with a positive genetic test result, a strong family history of cancer (regardless of their test result), or for people who have not had cancer and are having predictive genetic testing.

MBS 73295

Detection of germline BRCA1 or BRCA2 gene mutations, in a patient with platinum-sensitive relapsed ovarian, fallopian tube or primary peritoneal cancer with high grade serous features or a high grade serous component, and who has responded to subsequent platinum-based chemotherapy, requested by a specialist or consultant physician, to determine whether the eligibility criteria for olaparib under the Pharmaceutical Benefits Scheme are fulfilled.

MBS 73296

*Characterisation of germline gene mutations, requested by a specialist or consultant physician, including copy number variation in BRCA1 and BRCA2 genes and one or more of the following genes STK11, PTEN, CDH1, PALB2, or TP53 in a patient with breast or ovarian cancer for whom clinical and family history criteria, as assessed by the specialist or consultant physician who requests the service using a quantitative algorithm, place the patient at **>10% risk** of having a pathogenic mutation identified in one or more of the genes specified above.*

Quantitative algorithms for calculating BRCA1 and BRCA2 risk:

- **Manchester Scoring System**
Evans DG, et al. J Med Genet 2017;0:1–8. doi:10.1136/jmedgenet-2017-104584
Free full-text PDF at <http://jmg.bmj.com/>. Development of an app TBA.
- **BOADICEA** (Centre for Cancer Genetic Epidemiology, University of Cambridge)
Lee et al. British Journal of Cancer (2014) 110, 535–545.
Web application <http://ccge.medschl.cam.ac.uk/boadicea/>
- **BRCAPRO** (BayesMendel, Department of Biostatistics and Computational Biology, Dana-Farber Cancer Institute, Boston, Massachusetts)
Software from <http://bcb.dfci.harvard.edu/bayesmendel/software.php>

MBS 73297

Characterisation of germline gene mutations, requested by a specialist or consultant physician, including copy number variation in BRCA1 and BRCA2 genes and one or more of the following genes STK11, PTEN, CDH1, PALB2, or TP53 in a patient who is a biological relative of a patient who has had a pathogenic mutation identified in one or more of the genes specified above, and has not previously received a service under item 73296.

MBS item 73297 covers predictive testing in unaffected individuals. The National Pathology Accreditation Advisory Council (NPAAC) sets standards for medical testing laboratories in Australia. NPAAC classifies predictive genetic tests as Level 2 genetic tests requiring professional genetic counselling to precede and accompany the test.

Other genes

In addition to BRCA1 and BRCA2, the following genes can also be included. There are other cancer risk genes not yet included in any of the MBS items. Careful selection and correlation with clinical and family history reduces the potential for identifying variants of uncertain significance.

STK11: A fault in the STK11 gene causes Peutz-Jeghers syndrome (PJS), which is associated with multiple hamartomatous polyps of the gastrointestinal tract, and mucocutaneous pigmentation. Screening for gastrointestinal polyps begins in childhood. PJS is also associated with an increased lifetime risk of colorectal, pancreatic, lung and female breast cancer as well as cervical cancer and sex cord tumours. A diagnosis of PJS can be made on clinical criteria alone, even in the absence of a genetic test. Individuals with suspected PJS may benefit from examination by a clinical geneticist.

PTEN: A fault in the PTEN gene causes Cowden syndrome, which is associated with multiple non-cancerous growths (hamartomas) and an increased risk of benign and malignant tumours, primarily of the breast, thyroid and endometrium. Affected individuals usually have a large head circumference and particular skin lesions, and they sometimes have intellectual disability. A diagnosis of Cowden syndrome can be made on clinical criteria alone, even in the absence of a PTEN mutation. Individuals with suspected Cowden syndrome may benefit from examination by a dermatologist and/or a clinical geneticist.

CDH1: A fault in the CDH1 (E-cadherin) gene causes Hereditary Diffuse Gastric Cancer (HDGC), which is associated with a strong susceptibility to diffuse gastric cancer, and lobular breast cancer in women.

PALB2 (truncating mutations only): Mono-allelic, loss-of-function mutations in PALB2 increase the risk of breast cancer two to six-fold. Other cancer risks and the implications of other types of mutations in PALB2 are unclear at present. Bi-allelic mutations cause a distinct condition (Fanconi anaemia).

TP53: A fault in the TP53 gene causes Li-Fraumeni syndrome (LFS), a very rare condition that is associated with an increased risk of numerous cancers – primarily breast cancer, sarcoma, brain cancer, leukaemia and adrenocortical carcinoma. Exposure to carcinogens and radiation is associated with an increased likelihood of cancer development, and should be avoided if possible. A diagnosis of LFS can be made based on clinical criteria alone, even in the absence of a TP53 mutation. Not all TP53 families meet the criteria for LFS. Effective surveillance for LFS is limited, beyond that which is available for breast cancer. Some LFS-associated cancers may develop during childhood.