RADIATION SENSITIVITY ASSAY

The Radiation Sensitivity Assay may help determine if you have the *TGFβ1* genetic variation associated with a low or high risk of developing fibrosis. This information may be helpful when deciding whether to give a tumour bed boost after whole-breast radiation, as a boost increases the risk of fibrosis while modestly decreasing recurrence risk. [1]

What is Radiation-Induced Fibrosis?

Radiation-induced fibrosis (RIF) is a long-term side effect of external beam radiation therapy for the treatment of cancer. [2] RIF usually appears 4 – 12 months after radiation therapy and may progress over several years. [2] The clinical presentation depends on the type of tissue exposed to irradiation. The development of RIF can be described as a "wound-healing response gone wrong", and may manifest as skin induration and thickening, muscle shortening and atrophy, limited joint mobility, lymphedema, mucosal fibrosis, ulceration, fistula, hollow organ stenosis and pain – all of which can significantly impact a patient's quality of life. [2,3]

What causes Radiation-Induced Fibrosis?

There have been several studies that suggest the radiosensitivity of the breast is determined by genetic factors. [1, 4-6] The C−509T *TGFβ1* variant allele may be used as a genetic marker to identify patients at elevated risk for fibrosis following radiotherapy. [1]

Transforming growth factor-b1 (*TGFβ1*) is the major cytokine responsible for the regulation of fibroblast proliferation and differentiation. [6] Differentiated fibroblasts synthesise collagens and proteoglycans in the extracellular matrix, and it has been suggested that an increase in these fibroblasts may trigger the development of fibrosis. [6]

Radiation induces long-term *TGFβ1* overexpression due to oxidative stress and an inflammatory response. [6] Elevated serum *TGFβ1* levels were correlated with an increased risk of fibrosis in breast and lung cancer patients, and a comparison of the genotypes of unaffected and affected patients has been genetically associated with variants in the *TGFβ1* gene. [6]

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Why would I consider Radiation Sensitivity testing?

Genomic Testing can determine if you have the *TGFβ1* genetic variation which may be associated with low or high risk of developing fibrosis.

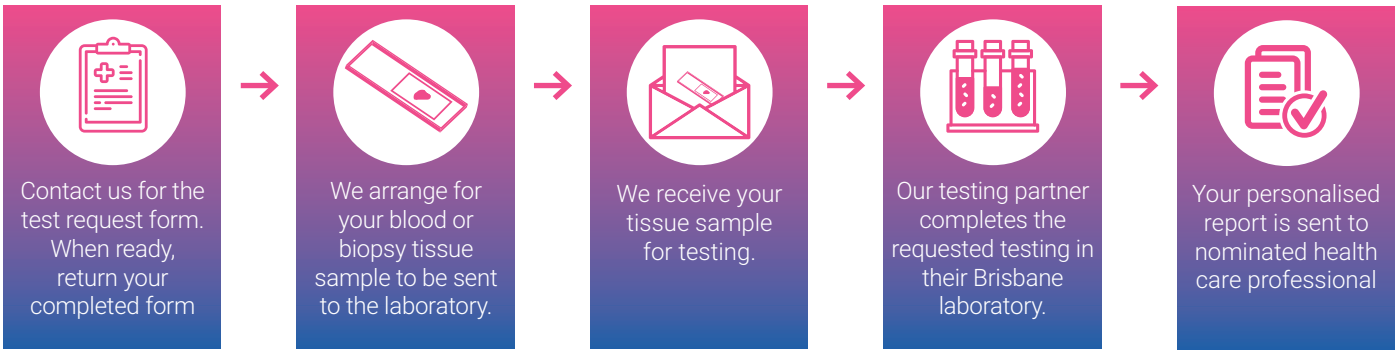
Around 50% of patients with solid malignant tumours receive radiation therapy at some point in the course of their disease. [7] For women with early-stage breast cancer, breast-conserving therapy involving breast-conserving surgery followed by whole-breast irradiation and optionally a boost to the tumour bed, is a standard therapeutic option. [8]

A boost to the tumour bed means that an extra dose of radiation is applied that covers the initial tumour site. A boost of radiotherapy to the tumour bed is commonly used as a local recurrence occurs mostly at the site of the primary tumour because remaining microscopic tumour cells are most likely situated there; and radiation can eliminate these microscopic tumour cells.[8]

Knowing whether you may have a low or high risk of developing fibrosis can be helpful when deciding whether to give a tumour bed boost after whole-breast radiation, as a boost may increase the risk of fibrosis while modestly decreasing recurrence risk. [1]

How do I organise Testing?

CG Genomics Oncology aims to educate patients and their families on their cancer type and empower them with the knowledge to take control of their treatment plans. As each patient's case is unique, there is no "one size fits all" when it comes to testing. We encourage you to contact CG Genomics Oncology, and we can work with you and your oncologist/ specialist, to determine what tests would benefit you.



Technical Information

The assay detects the *TGFB1* SNP ID rs1800469 (NM_030578.3): c.*309T>C (C-509T/c.-509C>T/c.-1347C>T) variant. All PCR based molecular diagnostic tests are subject to a low risk of non-amplification due to primer binding site variants which can lead to a false negative result.

References

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