

Diagnostic Assay for Gastrointestinal stromal tumors (GIST)

Technology

GIST can occur anywhere along the GI tract, but most often are found in the stomach or small intestine. Patients with GIST are treated by surgery and/or medical treatment with tyrosine kinase inhibitors. Response to therapy is evaluated by diagnostic imaging, which displays very limited sensitivity and specificity. To this date there is no biomarker available for detecting success or failure of therapy.

Activating mutations of CKIT or PDGFRA are hallmarks of 90% of GIST cases, where CKIT or PDGFR α DNA fragments are released into the circulation. Allele-specific PCR is used to specifically amplify and quantify mutated CKIT and PDGFR DNA fragments, which are highly specific for the disease.

Results of a prospective, explorative trial for the detection of circulating cell-free tumor DNA in the plasma of patients with GIST indicates that cell-free tumor DNA in the plasma can be used as tumor-specific biomarker.

Innovation

Detection of resistance mutations in plasma samples:

- earlier diagnosis and higher sensitivity
 - obviates the need for repeated tumor biopsies and imaging
 - allow earlier treatment changes may or may enable patients to take “drug holidays”
- ⇒ better treatment

Application

- Diagnosis of early stage GIST
- Therapy monitoring of GIST

Developmental Status

Assay is ready for scale up; little development work to be done

Responsible Scientist

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Patent Status

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