# Tumor Samples Harboring Novel Stem Cell Factor FOXD3 Variants

**Summary (1024-character limit)**
The National Institute of Child Health and Human Development Endocrinology & Genetics Section seeks parties interested in licensing tumor samples harboring novel stem cell factor FOXD3 variants.

**NIH Reference Number**
E-058-2010

**Product Type**
- Research Tools

**Keywords**
- Gastrointestinal Tumors (GIST)
- Paraganglioma Tumors
- FOXD3

**Collaboration Opportunity**
This invention is available for licensing.

**Description of Technology**

GISTs are one of the most common sarcomas of the gastrointestinal tract with an estimated 5,000-10,000 new cases in the U.S. reported each year. GISTs affect mainly pediatric and young adult patients, and respond poorly to current therapies. Paragangliomas are rare neuroendocrine neoplasms that develop primarily in the abdomen.

Researchers at the National Institute of Child Health and Human Development (NICHD),Section on Endocrinology and Genetics have made available samples of patient-derived gastrointestinal tumors (GIST) and paraganglioma tumors that harbor genetic mutations that have an effect on early stage embrogenesis, which plays a role in the fate of stem cells.

The tumor samples made available herein contain deletions in the FOXD3 gene and display down-regulated FOXD3 protein expression. While the majority of GISTs result from activating mutations in the oncogene receptor tyrosine kinases c-KIT and PDGFRA, these tumor samples do not harbor mutations in c-KIT or PDGFRA ("non-KIT/ PDGFRA-GISTs") and respond poorly to receptor tyrosine kinase inhibitors.

**Potential Commercial Applications**

- Useful in the investigation of inactivating genetic changes in FOXD3 in non-KIT/ PDGFRA-GISTs.
- Useful in the study of new molecules and/ or pathways that may serve as an appropriate therapeutic target.

**Inventor(s)**

Constantine A. Stratakis M.D, D.Sc. (NICHD)

**Development Stage**

- Prototype

**Patent Status**

- **Research Material:** NIH will not pursue patent prosecution for this technology

**Therapeutic Area**

- Gastrointestinal