Cancer Break Out Group

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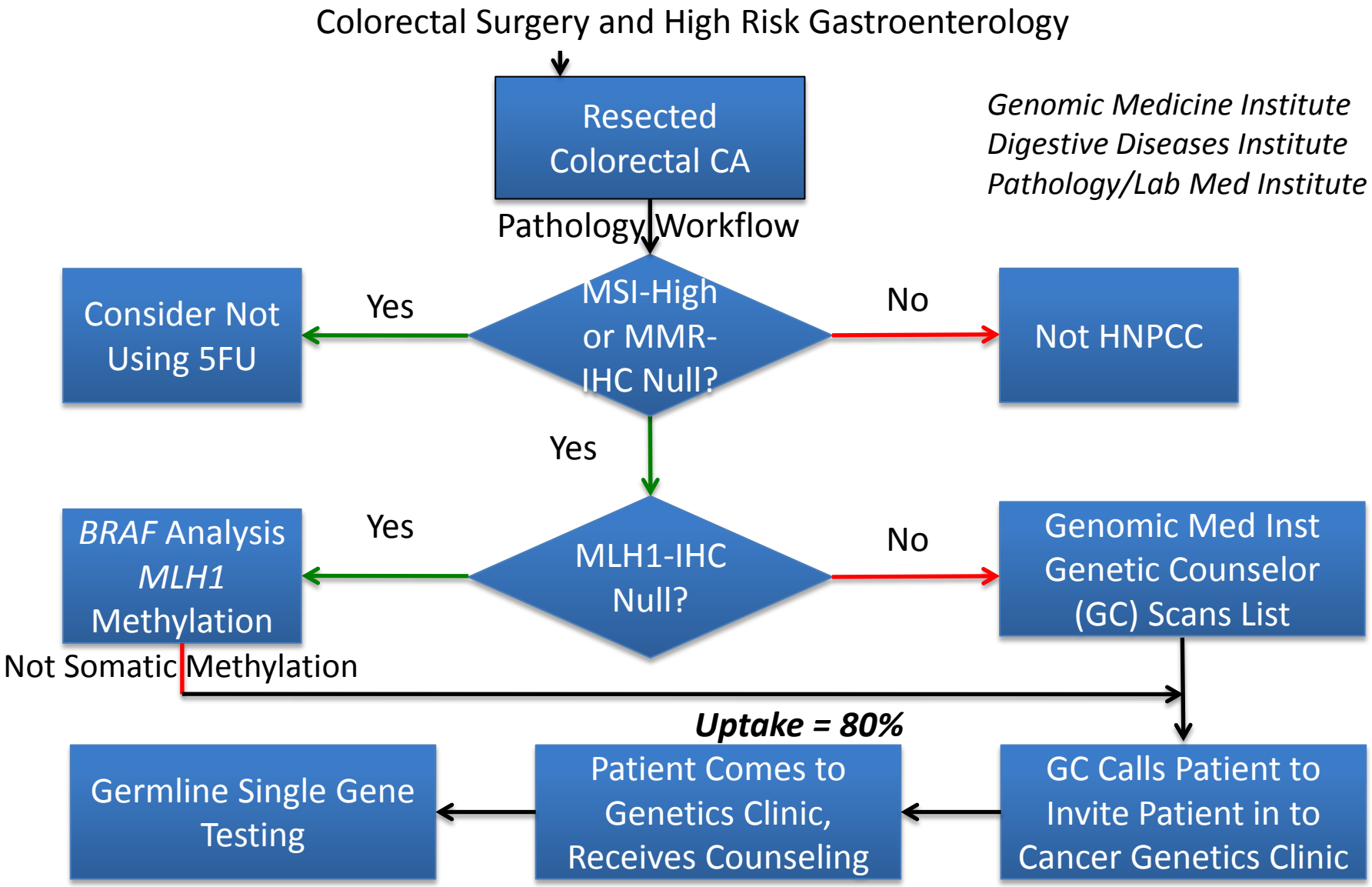
Lynch Syndrome Screening

- Goals
  - Improve implementation of recommendations for IHC/MSI screening for colorectal and endometrial cancers
  - Create a resource to evaluate successful implementation of screening
  - Aggregate/Integrate germline and tumor sequencing with treatment and outcomes including family pairs in order to understand variable penetrance, expressivity, and clinical outcomes

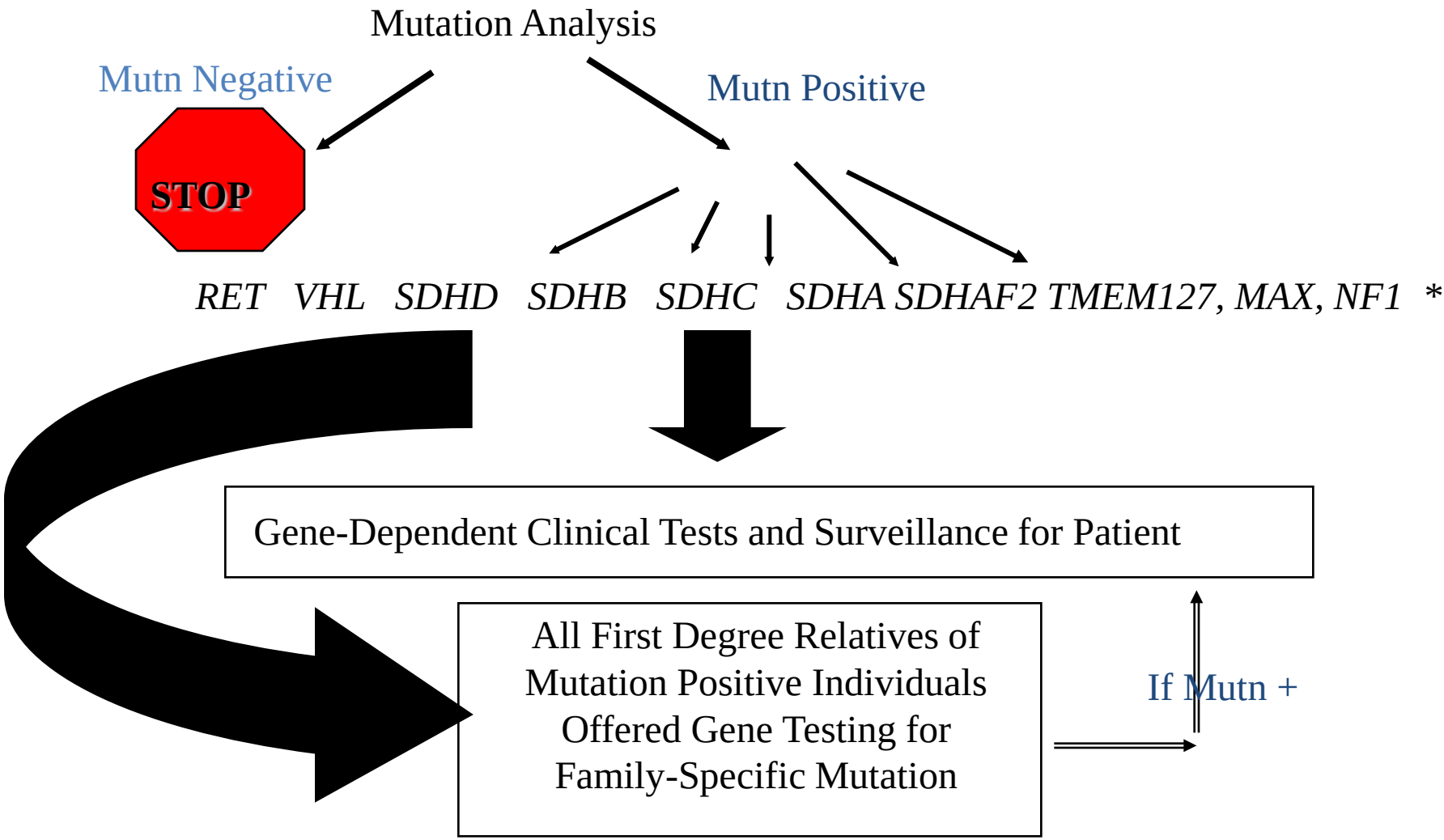
Cleveland Clinic Clinical Workflow for Screening All CRC for Lynch Syndrome (2009-onwards)



Neuroendocrine Cancer Screening

- Goals
  - Improve implementation of routine genetic screening for medullary thyroid carcinoma and pheochromocytoma/paraganglioma for every person
  - Create a resource to evaluate successful implementation of screening
  - Link to family history and TCGA projects

All Pheochromocytoma/Paraganglioma Patients Routinely to Germline Testing



*Capture and Targeted Resequencing vs Whole Genome Sequencing and Targeted Analysis?

Important Crumbs Left Behind

- Moderate risk variants
  - Clinical utility
  - Screening and treatment recommendations
- Very rare (and probably genetic) phenotypes with no known associated genes
- Germline and somatic variation for tumor progression and drug resistance
- Cancers that rarely have somatic alterations
  - Carcinoids, pancreatic endocrine tumors