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MAYO CLINIC LABORATORIES

Primary Ciliary Dyskinesia Genetic Testing Patient Information

Instructions: Accurate interpretation and reporting of genetic results is contingent upon the reason for testing, clinical information, ethnic background/ancestry, and family history. To help provide the best possible service, supply the information requested below and **send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Molecular Technologies Laboratory Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: +1-507-266-5700 or email MLIINT@mayo.edu**

Patient Information

Patient Name (Last, First Middle)	Birth Date (mm-dd-yyyy)

Sex Assigned at Birth	Legal/Administrative Sex
☐ Male ☐ Female ☐ Unknown ☐ Choose not to disclose	☐ Male ☐ Female ☐ Nonbinary

Referring Healthcare Professional Information

Requesting Healthcare Professional Name (Last, First)	Phone	Fax*
Genetic Counselor/Other Healthcare Professional Name (Last, First)	**Phone**	**Fax***

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Reason for Testing

☐ Diagnosis ☐ Family History** ☐ Other, specify:

**Genetic testing should be performed on an affected family member first, when possible. FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family.

Clinical History

Clinical Findings

☐ Situs abnormality
- ☐ Situs inversus totality
- ☐ Heterotaxy
- ☐ Dextrocardia/Congenital heart defect
- ☐ Asplenia/Polysplenia
- ☐ Pulmonary isomerism
- ☐ **Other, specify:**

☐ Chronic nasal congestion

☐ Chronic sinusitis

☐ Pulmonary disease
- ☐ Neonatal respiratory distress
- ☐ Chronic airway infections
- ☐ Bronchiectasis
- ☐ Pulmonary calcium deposits

☐ Chronic or recurrent ear infections

☐ Infertility

Laboratory Findings

☐ Abnormal ciliary ultrastructure
- ☐ Shortening/Absence of outer dynein arms
- ☐ Shortening/Absence of both outer and inner dynein arms
- ☐ Microtubular disorganization
- ☐ Absence/Disruption of the central apparatus
- ☐ Other, specify:

☐ Abnormal ciliary motility

☐ Low nasal nitric oxide: nl/min

Other Relevant Clinical History

Family History

Are there similarly affected relatives? ☐ Yes ☐ No
If "Yes," indicate relationship and symptoms:

Have any family member had genetic testing? ☐ Yes*** ☐ No ☐ Unknown
***FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family. Contact the lab for ordering assistance.**

History of consanguinity: ☐ No ☐ Yes; relationship details:

Ancestry

☐ African/African American	☐ East Asian	☐ Latinx/Latine	☐ South Asian	☐ Unknown
☐ Ashkenazi Jewish	☐ European	☐ Middle Eastern	☐ None of the above	☐ Choose not to disclose

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576) or Informed Consent for Genetic Testing – Spanish (T826).

Print