



**FORM 1- Instructions for submission of specimen for DNA testing**

**The patient should be fully informed about the test.**

**Nature of the test:** The test detects mutations in gene(s) involved in the synthesis of proteins of connective tissue. Extraction of DNA from blood or tissue of the affected individual is required.

**Test limitations:** Only the gene implicated in the disorder will be studied which your physician will determine. Mutations in other genes will not be detected. The rate of mutation detection varies with the disorder and the gene studied. Mutations in non-structural portions of the gene will generally not be detected.

**Test results:** The test results are reported to the physician in writing. The test generally takes about 2-4 weeks; we try to expedite the analysis for prenatal testing.

**Sample requirements**

•**Whole Blood:** 10 cc (adults) in purple top (EDTA) tube should be drawn by venipuncture. In the case of small children, 3-5cc of EDTA blood is generally sufficient amount to complete the analysis. There are no special dietary or blood drawing considerations. Patient must not have received a transfusion recently. Heparin inhibits the PCR reaction, therefore heparin tubes are not acceptable. DNA extracted from heparin-contaminated blood is not accepted

•**Fibroblasts, amniocytes, CVS:** Preferably 2-4 confluent T25-flasks of cultured cells. It is highly advisable to call prior to submitting cultured cells.

•**Tissues:** Please call the lab at 504-988-7706. We do not take whole tissue or paraffin blocks. We do accept extracted DNA from tissues, but not from paraffin blocks.

**Sample submission**

Sample should be shipped by **overnight courier, preferably Federal Express** (Use DHL/Airborne with caution as we have experienced significant delivery issues with them). Please send with a cold pack during summer months. If blood sample can't be shipped the same day, please store at +4C until shipment. All tubes/flasks should be labeled with patient name and date of birth, or other identifying criteria. Please do not send any samples on Friday. **Patient Information Form** (Form 2), **Payment Information Form** (Form 3) and an **OPTIONAL signed Patient Consent Form** should accompany the sample. If laboratory fee is paid by check that accompanies the sample, Form 3 is not needed. Check should be made out to **Tulane University**. We can assist with test description and related information for insurance companies.

<b>Charges</b>	
<b>Osteogenesis Imperfecta:</b> Collagen I Gene (COL1A1 and COL1A2) Analysis	\$1800
<b>Chondrodysplasias:</b> Collagen II Gene (COL2A1) Analysis	\$1400
<b>Stickler/Marshall Syndrome:</b> Analysis of the COL2A1 and COL11A1 Genes	\$2300
<b>Marfan Syndrome:</b> Fibrillin -1 gene (FBN1) Analysis	\$1400
<b>Metaphyseal dysplasia type Schmid:</b> Collagen X gene (COL10A1) Analysis	\$460
<b>Known mutation (family member or prenatal)</b> in any of the above	\$350

**Sample and Paperwork should be sent to:** Please call (504) 988-7706 before submitting a sample for the first time **if you need assistance.**

Matrix DNA Diagnostics  
 Attn: DNA Diagnostics Laboratory  
 Center for Gene Therapy  
 Tulane University Health Science Center  
 1430 Tulane Avenue, TB 28  
 Tidewater 2140  
 New Orleans, LA 70112



<b>Patient name:</b>	<b>Date of Birth:</b>
<b>Hospital Reference Number:</b>	<b>SSN:</b>
<b>Address:</b>	<b>Telephone:</b>

**Patient Diagnosis/Clinical information:**

**Required attachments:**

- copy of a driving license for the policy holder, if insurance bill
- copy of clinical history and physical examination

<b>Check for the test requested:</b>	<input type="checkbox"/>	COL1A1/COL1A2 gene analysis for Osteogenesis Imperfecta
	<input type="checkbox"/>	COL2A1 gene analysis for SED, ACGII, HCG, Kniest dysplasia
	<input type="checkbox"/>	COL2A1 /COL11A1 gene analysis for Stickler/Marshall Syndrome
	<input type="checkbox"/>	FBN1 gene analysis for Marfan Syndrome
	<input type="checkbox"/>	COL10A1 gene analysis for Schmid Metaphyseal dysplasia
Proband name must be provided in box above.	<input type="checkbox"/>	Known Mutation (family member or prenatal) in any of the above

<b>Type of specimen:</b>	<b>Is this a prenatal test YES NO</b>
<b>Collected by:</b>	<b>Collection date:</b>

<b>Referring Physician Name</b>	<b>Contact person</b>
<b>Telephone Fax</b>	<b>Address</b>
<b>Request Date</b>	<b>Physician's Authorized Signature</b>

**If this is a known family mutation, please provide the proband's name or the proband report.**

### Form 3: Payment Information/Invoice

**1. Payment by check.** Please make the check payable to **Tulane University** and send to:

Matrix DNA Diagnostics  
Center for Gene Therapy  
1430 Tulane Ave, SL-99  
New Orleans, LA 70112

<b>ICD-9 code:</b>	756.5	<b>Tax/Fed ID#:</b>	72-0423889	<b>CLIA#:</b>	39D0903989
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**2. Payment by Institution.**

Contact Person:	Phone number	Bill to- address
	Fax number	

**3. Payment by Insurance Company. ATTACH A READABLEPHOTOCOPY OF THE INSURANCE CARD (BOTH SIDES)! CARDHOLDER INFORMATION MUST ALSO BE INCLUDED (address and date of birth)!**

Name of Policy Holder:	SSN or member#:	Plan/Group#:
Insurance Provider's Name:	Phone Number:	Address:
Claim Dept's phone #:	Insurance Contact person:	Pre-approval number:

**DISCLAIMER:** DNA Diagnostics may not be an in-network provider with most insurance companies. We reserve the right to decline/refuse any insurance as a payment option. The patient accepts full responsibility for any payment not covered by insurance. **Patients may be required to provide alternate funding, upon request, in advance for any amount that may not be covered by insurance. WE DO NOT ACCEPT ANY MEDICAID.**

#### Known Familial Mutation Analysis

CPT codes	Description	# Performed	Cost/test	Extended
83890	DNA isolation	1	\$10.00	\$10.00
83898	Polymerase chain reaction	6	\$15.00	\$ 90.00
83904	Nucleic acid probe w/ sequencing	6	\$40.00	\$ 240.00
83912	Interpretation and report	1	\$10.00	\$10.00
<b>TOTAL COST</b>				<b>\$ 350.00</b>

\*Please note that the Family Known Mutation analysis is a multistep test, which involves sequencing of a portion of a very large gene and, therefore, involves multiple CPT codes. For more information, please call the lab about the test (504-988-7706)

<b>In signing this form, I indicate that I understand the information presented above and agree to these stipulations.</b>	
<b>Patient Signature</b>	<b>Date</b>