CENTRES OFFERING GENE TESTING SERVICES FOR GENETIC DISORDERS

INDIAN ASSOCIATION OF DERMATOLOGISTS, VENEREOLOGISTS AND LEPROLOGISTS

- NORTH ZONE
- WEST ZONE
- SOUTH ZONE
- OTHER CENTRES
- WORLDWIDE CENTRES

Disclaimer:
This may not be an exhaustive list of centers offering testing for genodermatoses and IADVL does not endorse any of the centers and will not be liable for any deficiency of services. The aim is to provide information for where testing may be available in each zone. Please refer to the individual website for more details.

Creative Partner
SIGMA Healthcare Communications

Besides offering highly sensitive **Diagnostic Testing**, the centers offers **Carrier Testing** (for family members), **Prenatal Testing** (to know if the new-born will be normal), **Susceptibility Testing** (to know if one is at risk), **Predictive Testing** (to inform chances of developing the condition later in life), and **Personalised Testing** (to know a person's individual response to certain life saving drugs).

The centers have been divided zone wise.

NORTH ZONE

SIR GANGA RAM HOSPITAL, CENTER OF MEDICAL GENETICS, NEW DELHI

ADDRESS

Rajinder Nagar, New Delhi.

PHONE

+91-1142251997

CONTACT PERSONS

- **Dr. Renu Saxena:** +91-1142252139, renu2006@gmail.com
- **Sudha Kohli:** +91-1142252114, kohli_sudha@yahoo.com
- **Ratna D Puri:** +91-1142251996, dr_icverma@yahoo.com

FACILITIES AVAILABLE

Genetic testing for following genodermatosis is available:

- Neurofibromatosis 1 Test - linkage analysis
- Oculocutaneous Albinism Type 1 Test - linkage analysis and genotyping
- Oculocutaneous Albinism Type 2 Test - linkage analysis and genotyping
- 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia Test
- Acute Intermittent Porphyria Test
- Alkaptonuria Test
- Ataxia-Telangiectasia Test
- Hypohidrotic Ectodermal Dysplasia, X-Linked Test
- Ichthyosis, X-Linked Test
- Incontinentia Pigmenti Test

CENTOGENE INDIA PVT LTD (THE RARE DISEASE COMPANY)

ADDRESS

107 Wegman's Business Park,
Knowledge Park III,
Surajpur-Kasna Road,
Greater Noida - 201308,
Uttar Pradesh.

PHONE

+91-8527317888

FAX

+91-1166173749

E-MAIL

india@centogene.com

WEBSITE

www.centogene.com

CONTACT PERSON

- **Dr. Sunil Tadepalli**
- **Phone:** +91-9910017978
- **Email:** sunil.tadepalli@gmail.com

FACILITIES AVAILABLE

CENTOGENE is one of the leading laboratories focusing on genetic testing of rare Hereditary Disorders. We now offer more than 2200 routine genetic and biochemical tests. In addition we perform analysis for biomarkers for lysosomal storage diseases. Centogene works with academic and industrial partners to develop cutting-edge diagnostic techniques and new orphan drugs. We have sample collection centers all over India.

DELHI DERMPATH LABORATORY

ADDRESS

10, Aradhana Enclave,
Sector - 13, R.K. Puram
New Delhi - 110066.

PHONE

+91-01124196635,
+91-01124196666,
+91-9810286755

CONTACT PERSON

- **Dr. Asha Kubba**
- **Phone:** +91-9810286755
- **Email:** delhidermpathlab@gmail.com

FACILITIES AVAILABLE

- Antigen mapping for Epidermolysis Bullosa (EB) group and variants

WEBSITE

www.kubbaskinclinic.com

SANJAY GANDHI POST GRADUATE INSTITUTE OF MEDICAL SCIENCES, LUCKNOW

SERVICES PROVIDED

The department conducts general outpatient clinic for genetic disorders thrice a week (Monday, Tuesday,Thursday).

PRENATAL DIAGNOSIS

Available for:

- Congenital malformations and multiple malformations syndrome
- Chromosomal disorders

HLA INVESTIGATIONS

- HLA Typing (class 1 and 2)
- HLA B27 typing

ALL NDIA INSTITUTE OF MEDICAL SCIENCES, NEW DELHI

FACULTY & STAFF (PERMANENT)

- **Dr. Madhulika Kabra**
  Additional Professor & Officer-in-Charge, Genetics Unit, Department of Pediatrics, AIIMS.
- **Email:** madhulikakabra@hotmail.com, mkabra_aiims@yahoo.co.in

SERVICES OFFERED

- Congenital adrenal hyperplasia. Testing for 6 common mutations and prenatal diagnosis.
- Oculo-cutaneous albinism. PCR based test for common mutations.
- Connexin 26 disorders.

WEST ZONE

GENEOMBIO TECHNOLOGIES, PUNE

ADDRESS

Vedant, S. No. 39/3,
H. No. 1043, Yogi Park,
Behind Periwinkle Building,
Off Mumbai Bangalore
Expressway, Baner,
Pune-411045.

PHONE

+91-9960000984
+91-9730073423
+91-020-30470652 / 53 / 54

E-MAIL

info@geneombiotechnologies.com,
helpdesk@geneombiotechnologies.com,
crs@geneombiotechnologies.com

WEBSITE

http://www.geneombiotechnologies.com

TESTS AVAILABLE

We specialize in providing Molecular Diagnostic Services to various healthcare organizations. Our core competency in Molecular Diagnosis lies in conducting Bacterial Pathogen PCR Test, Viral Pathogen PCR Test, Karyotyping Test, Fluorescent In Situ Hybridization Test, Torch Panel Test, Advanced Genome Analysis Test, Pharmacogenomics Test, Viral Load and Genotyping Test, Genetic Analysis PCR Test, Cancer Genetics Test, Predictive Genetic Test, Genodermatology Test, Aesthetic Genetics Test, and Human Congenital Disorder Test.

CENTRE FOR GENETIC DIAGNOSIS, DEENANATH MANGESHKAR HOSPITAL & RESEARCH CENTER, PUNE

ADDRESS

Near Mhatre Bridge,
Erandawne,
Pune 411004.

PHONE

+91-2040151000 /
66023000

FAX

+91-2025420104

E-MAIL

india@centogene.com

NAVI MUMBAI INSTITUTE OF RESEARCH IN MENTAL AND NEUROLOGICAL HANDICAP (NIRMAN), MUMBAI

ADDRESS

A - 103, Vardhaman
Chambers, Above Axis Bank,
Sector 17, Vashi,
Navi Mumbai, India - 400 705.

PHONE

+91-2267910236 / 37

CONTACT PERSONS

- **Dr. Anil B. Jalan**
  NIRMAN +91-9821124578, +91-2267910236
- **Mr. Sushil Upadhyay (Manager)**
  NIRMAN +91-9892158501, +91-2267910237

WEBSITE

www.metabolicerror.com

FACILITIES AVAILABLE

Genetic testing for following genodermatosis are available:

- Sjogren Larsson Syndrome (SLS) (ALDH3A2)
- Tuberous Sclerosis 1
- Ehlers-Danlos Syndrome, type VII
- Ataxia Telangiectasia
- Protein C Deficiency, Congenital (PROC)
- Protein S Deficiency, Congenital (PROS1)
- Refsum disease
- Hereditary sensory motor neuropathies
- Charcot-Marie Tooth Disease

SOUTH ZONE

CENTRE FOR CELLULAR AND MOLECULAR BIOLOGY, MOLECULAR DIAGNOSTICS DIVISION, HYDERABAD

ADDRESS
Uppal Road, Habshiguda
Hyderabad 500007.

PHONE
+91-4027192549

CONTACT PERSONS
- **K Radha Mani**
- **E-mail:** nidan@ccmb.res.in
- **Dr. Giriraj Ratan Chandak**
- **E-mail:** chandakgrc@ccmb.res.in
- **Phone:** +91-4027192748

TESTS AVAILABLE
- Glucose 6 phosphate dehydrogenase deficiency - Orissa, Mediterranean and Kerala-Kalyan mutations.
- Pre-coagulation profile - Factor V Leiden and Prothrombin (G20210A) mutation.

DNA LABS INDIA, HYDERABAD

ADDRESS
HNo: 3-6-151-153,
G S Towers, Flat No 303,
3rd Floor, Opposite Mithra
Agencies, Himayat Nagar,
Hyderabad - 500029, India.

PHONE
+91-04067119116

E-MAIL
hyderabad@dnalabsindia.com

Regional labs available in Delhi, Mumbai, Kolkata, Bangalore, Ahmedabad.

WEBSITE
www.dnalabsindia.com

FACILITIES AVAILABLE
- Epidermolysis bullosa panel (NGS Panel; COL17A1, LAMA3, LAMB3, LAMC2) - 45,000/-
- Genetic Ehlers-Danlos syndrome panel NGS Panel COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, TNXB DNA Test - 45000/-
- Genetic Refsum disease panel NGS Panel: PEX1, PEX2, PEX26, PEX7, PHYH DNA Test - 45000/-
- Genetic Mucopolysaccharidosis panel NGS Panel: IDUA, IDS, SGSH, NAGLU, HGSNAT, GNS, GALNS, ARSB, GUSB, HYAL1, LDB3, MYOT DNA Test - 45000/-
- HLA - B27 - 3,800/-
- DNA PCR testing for HPV, CMV, HBV also available.

KIMS FOUNDATION AND RESEARCH CENTER (KFRC), SECUNDARABAD

ADDRESS
KIMS Foundation and
Research Center (KFRC)
#1-8-31/1, Minister Road,
Secunderabad - 500 003. A.P., India.

PHONE
+91-4044885170

FAX
+91-4027840980

E-MAIL
kfrc2010@gmail.com

WEBSITE
www.kfrc.co.in

CENTER FOR HUMAN GENETICS, BENGALURU

ADDRESS
Biotech Park, Electronic City Phase I,
Bengaluru - 560 100, Karnataka, India.

PHONE
+91-8028521382 / 28521833

FAX
+91-8028521832

GENETECH PRIVATE LIMITED, HYDERABAD

ADDRESS
6-3-1113/4, Greenlands, Hyderabad
Andhra Pradesh, INDIA - 500 016.

PHONE
+91-4023400932,
23406178, 55638754

E-MAIL
info@genetech.co.in

FACILITIES
Gene analysis for albinism

FAX
+91-4023407943

SERVICES
Medical Genetics / Clinical Genetics OPD Consultations:
- Monday - Genetic Counselling Clinic 2-4pm at Clinical Genetics unit.
- Tuesday - Genetic Counselling Clinic 2-4 pm at Clinical Genetics unit.
- Wednesday / Saturday - Medical Genetics OPD 8 am - 12 noon at OPD building 250, room no 4,5,6.
- Wednesday (Multispecialty) Perinatal Medicine Clinic 2.30 - 4 pm at ISSC BUILDING First floor, room no 3.
- Thursday (Multispecialty) Neuromuscular Clinic 2 - 4.30 pm at PMR building room no. 9.

Molecular tests available for common single gene disorders such as:

1. Achondroplasia, Duchenne muscular dystrophy / Becker muscular dystrophy, Spinal muscular atrophy, Methylation testing for Angelman/Prader Willi, **Connexin26**, Huntington disease, Myotonic dystrophy, Spinocerebellar Ataxia (SCA 1,2,3, and 7), **Neurofibromatosis , Ehlers-Danlos syndrome, Alkaptonuria, Gaucher disease, Mucopolysaccharidosis,** Metabolic and inborn errors such as homocystinuria etc.
2. Prenatal testing of the above conditions if indicated.
3. Presymptomatic diagnosis for late onset genetic disorders when indicated.

GENES N LIFE HEALTH CARE PVT. LTD., HYDERABAD

CONTACT DETAILS
D.No. 6-3-647/9, F. No. 204
Riviera Buildings,
Dwarakapuri Colony Punjagutta,
Hyderabad - 500 082.

PHONE
+91-04023350551

FAX
+91-04023350330

E-MAIL
info@genesnlife.com; genesnlife@gmail.com

WEBSITE
http://www.genesnlife.com/

OTHER CENTRES

OTHER CENTRES WHICH ARE OFFERING GENETIC TESTING FACILITIES IN INDIA ARE:

- Kokilaben Dhirubhai Ambani hospital and medical research institute, Andheri, Mumbai.
- Hinduja Hospital, Mumbai.
- PSG hospital, Coimbatore.
- Genetics Cell, Sri Ramachandra Medical College (SRMC) Porur, Chennai.
- Genetics department, Manipal Hospital, Karnataka.

However, a lot of genetic tests are not available in India. Here is a list of some of the rare tests available worldwide which are of relevance to a dermatologist.

WORLDWIDE CENTRES

ACRODERMATITIS ENTEROPATHICA (SEQUENCE ANALYSIS OF SLC39A4 GENE)

- CGC Genetics - *Porto, Portugal*
- Reference Laboratory Genetics - *Barcelona, Spain*
- Cincinnati Children's Hospital Medical Center, Molecular Genetics Laboratory - *Cincinnati, OH, USA*
- Sistemas Genomicos, Medical Genetics Unit - *Paterna, Spain*
- Medical Genetics Center, Asian Medical Center - *Seoul, Korea*

ACUTE INTERMITTENT PORPHYRIA, SEQUENCING HMBS GENE

- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*
- Center for Human Genetics and Laboratory Medicine Martinsried, Molecular Genetics - *Martinsried, Germany*
- Reference Laboratory Genetics - *Barcelona, Spain*
- Mayo Clinic - Minnesota, Molecular Genetics Laboratory - *Rochester, MN, USA*

ACUTE PORPHYRIA, MULTI-GENE PANEL

- Mayo Clinic - Minnesota, Molecular Genetics Laboratory - *Rochester, MN, USA*
- Center for Human Genetics and Laboratory Medicine Martinsried, Molecular Genetics - *Martinsried, Germany*

ALOPECIA UNIVERSALIS (SEQUENCE ANALYSIS OF HR GENE)

- Reference Laboratory Genetics - *Barcelona, Spain*
- CGC Genetics - *Porto, Portugal*

AUTOSOMAL RECESSIVE CUTIS LAXA TYPE IA, SEQUENCING FBLN5 GENE

- Reference Laboratory Genetics - *Barcelona, Spain*

AUTOSOMAL RECESSIVE CUTIS LAXA TYPE IC, SEQUENCING LTBP4 GENE

- Reference Laboratory Genetics - *Barcelona, Spain*

AUTOSOMAL RECESSIVE CUTIS LAXA TYPE IIA, SEQUENCING ATP6V0A2 GENE

- Reference Laboratory Genetics - *Barcelona, Spain*
- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*

AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI) VIA THE ABCA12 GENE

- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*

AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI) VIA THE TGM1 GENE

- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*

BENIGN CHRONIC PEMPHIGUS TEST

- Medgene, MedGene - *Bratislava, Slovakia*
- Praxis fuer Humangenetik Wien - *Vienna, Austria*

BLOOM SYNDROME (BLM) 1 MUTATION

- ARUP Laboratories, Molecular Genetics Laboratory - *Salt Lake City, UT, USA*
- Mayo Clinic - Minnesota, Molecular Genetics Laboratory - *Rochester, MN, USA*
- Recombine - *Livingston, NJ, USA*
- Sistemas Genomicos, Medical Genetics Unit - *Paterna, Spain*
- Hospital for Sick Children, Genome Diagnostics Laboratory (formerly Molecular Genetics Laboratory) - *Toronto, Canada*

BROOKE-SPIEGLER SYNDROME (SEQUENCE ANALYSIS OF CYLD GENE)

- CGC Genetics - *Porto, Portugal*
- GENETAQ, Molecular Genetics Centre - *Malaga, Spain*
- Reference Laboratory Genetics - *Barcelona, Spain*

BUSCHKE-OLLENDORFF SYNDROME (BOS)

- Connective Tissue Gene Tests - *Allentown, PA, USA*
- Centogene AG, Rare Disease Company - *Rostock, Germany*
- Innovagenomics S.L, Innovagenomics - *Salamanca, Spain*

CHEDIAK-HIGASHI SYNDROME (SEQUENCE ANALYSIS OF LYST GENE)

- CGC Genetics - *Porto, Portugal*
- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*
- Casey Molecular Diagnostic Laboratory, CEI, OHSU - *Portland, OR, USA*
- Reference Laboratory Genetics - *Barcelona, Spain*
- Cincinnati Children's Hospital Medical Center, Molecular Genetics Laboratory - *Cincinnati, OH, USA*

COCKAYNE SYNDROME A (SEQUENCE ANALYSIS OF ERCC8 GENE) & COCKAYNE SYNDROME B (SEQUENCE ANALYSIS OF ERCC6 GENE)

- CGC Genetics - *Porto, Portugal*

COL17A1-RELATED JUNCTIONAL EPIDERMOLYSIS BULLOSA TEST

- Centogene AG, Rare Disease Company - *Rostock, Germany*
- Connective Tissue Gene Tests - *Allentown, PA, USA*

EPIDERMOLYSIS BULLOSA SIMPLEX (SEQUENCE ANALYSIS OF KRT14 GENE) & EPIDERMOLYSIS BULLOSA SIMPLEX (SEQUENCE ANALYSIS OF KRT5 GENE)

- CGC Genetics - *Porto, Portugal*
- Cincinnati Children's Hospital Medical Center, Molecular Genetics Laboratory - *Cincinnati, OH, USA*
- Instituto de Medicina Genómica, IMEGEN - *Paterna (Valencia), Spain*
- Centogene AG, Rare Disease Company - *Rostock, Germany*

EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL DOMINANT (DDEB) & EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE (RDEB)

- Connective Tissue Gene Tests - *Allentown, PA, USA*

EPIDERMOLYSIS BULLOSA JUNCTIONAL (SEQUENCE ANALYSIS OF COL17A1 GENE) & EPIDERMOLYSIS BULLOSA JUNCTIONAL (SEQUENCE ANALYSIS OF LAMA3, LAMB3, LAMBC2 GENE)

- CGC Genetics - *Porto, Portugal*
- Cincinnati Children's Hospital Medical Center, Molecular Genetics Laboratory - *Cincinnati, OH, USA*

ECTODERMAL DYSPLASIA 1, HYPOHIDROTIC, X-LINKED (XHED)

- Connective Tissue Gene Tests - *Allentown, PA, USA*

EPIDERMODYSPLASIA VERRUCIFORMIS, SEQUENCING TMC6 and TMC8 GENE

- Reference Laboratory Genetics - *Barcelona, Spain*

ERYTHROKERATODERMIA VARIABILIS (SEQUENCE ANALYSIS OF GJB3 GENE)

- CGC Genetics - *Porto, Portugal*

ERYTHROPOIETIC PROTOPORPHYRIA, AUTOSOMAL RECESSIVE

- MVZ Dortmund Dr. Eberhard and Partner - *Dortmund, Germany*
- Diagenom GmbH, Medical Genetics Laboratory - *Rostock, Germany*
- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*
- CGC Genetics - *Porto, Portugal*

FABRY DISEASE TEST

- Centogene AG, Rare Disease Company - *Rostock, Germany*
- Mount Sinai School of Medicine, Mount Sinai Genetic Testing Laboratory (DNA Division) - *New York, NY, USA*
- Medgene, MedGene - *Bratislava, Slovakia*
- GGA - Galil Genetic Analysis - *Kazerin, Israel*

GORLIN SYNDROME, SEQUENCING PTCH1 GENE

- Reference Laboratory Genetics - *Barcelona, Spain*
- PreventionGenetics (Prevention Genetics), Clinical DNA Testing and DNA Banking - *Marshfield, WI, USA*
- CGC Genetics - *Porto, Portugal*

GOLTZ SYNDROME, SEQUENCING PORCN GENE

- Reference Laboratory Genetics - *Barcelona, Spain*
- Academic Medical Centre, University of Amsterdam, DNA Diagnostics Laboratory - *Amsterdam, Netherlands*

HAILEY-HAILEY DISEASE: ATP2C1 GENE SEQUENCE ANALYSIS

- GENETAQ, Molecular Genetics Centre - *Malaga, Spain*