Molecular Genetic Diagnosis
¿WHO WE ARE?

CIDEGEN is a molecular diagnostic center that provides comprehensive diagnostic and R&D services. Our aim is to facilitate and offer physicians and other specialists innovative and cutting edge cytogenetic and molecular testing, helping them to choose the right test to order and helping to interpret results in terms of diagnosis, prognosis and therapeutic relevance for their patients.

CIDEGEN was created in 2011 by a multidisciplinary team of specialists in molecular diagnostics, physicians, professors and researchers from different areas of clinical and biotechnological knowledge, with the determination to offer unique and high quality diagnostic services. As a partner company of the University of Salamanca and the CIC (Centro de Investigación del Cancer USAL-CSIC) CIDEGEN has experienced advice and support from renowned members of the scientific community, especially in the fields of genetic diagnostics and clinical research.

OBJECTIVES

CIDEGEN objectives is to promote and assist research projects and offer a focused genetic diagnosis analytical services, collaborating with National Health Systems to facilitate access to these techniques.
CIDEGEN headquarters has provided equipment and latest technology to perform the most modern techniques in molecular diagnosis. Our R&D lab, enables us to provide genetic services "à la carte", making possible the implementation by commissioning research on rare diseases and project collaboration at all levels and wherever possible.

All our activities are controlled by strict quality standards based on a system of risk analysis, identification and control of critical points. Our facilities have all regulatory approvals, protection of databases and security systems on information sent to our customers. Annually we pass strict quality controls and auditions by external entities.

In our desire to improve and offer the latest advances in clinical diagnosis, CIDEGEN collaborates with various clinical research groups and is part of research projects funded by the Spanish Ministerio de Economía y Competitividad.
CIDEGEN offers a wide range of genetic tests performed with the latest technologies and personalized genetic counseling in each analysis:

**Molecular cytogenetics** (Fluorescent in situ hybridization (FISH), micro-arrays)

**Molecular biology** (PCR, piro-sequencing, Next Generation Sequencing)

## GENETIC TESTS

### HEMATOLOGY/ONCOLOGY

- CML – Chronic Myeloid Leukemia
- AML – Acute Myeloid Leukemia
- CLL – Chronic Lymphoblastic Leukemia
- ALL – Acute Lymphoblastic Leukemia
- Myelodysplastic syndromes
- Multiple Myeloma
- Lymphoma
- Lymphoproliferative disorders
- Myeloproliferative disorders
- Thrombophilia
- Histocompatibility

### SOLID TUMORS

- Colorectal cancer
- Lung cancer
- Breast cancer
- Ovarian cancer
- Prostate cancer
- Bladder cancer
- Neuroblastoma
- Sarcoma
- Glioma
- Melanoma

### INHERITED DISEASES

- Hereditary cancer
- Neurodegenerative diseases
- Congenital cardiopathies
- Hereditary diseases

## REAGENTS

CIDEGEN, provides a great variety of products to meet the needs of flow cytometry laboratories under the brand CIDEGEN Reagents.

CIDEGEN Reagents offers a full range of laboratory solutions, including fluorochrome conjugated isotype controls and a wide range of assays, kits and reagents for standardization of instruments and sample preparation antibodies.

Request our catalog or download it from our web [www.cidegen.com](http://www.cidegen.com)
HEMATOLOGY/ONCOLOGY (CANCER & BLOOD DISEASES)

ACUTE MYELOID LEUKEMIA (AML)
- MLL, del(11q23)
- RARα, t(17q21.1)
- BCR/ABL, t(9;22)(q34;q11)
- Nucleophosmin NPM1 (5q35)
- FLT3 (13q12.2)

ACUTE MYELOID LEUKEMIA M2 (AML - M2)
- AML1/ETO, t(8;21)(q22;q22)

ACUTE PROMYELOCYTIC LEUKEMIA M3 (AML – M3)
- PML/RARα, t(15;17)(q22;q21)

ACUTE MYELOID LEUKEMIA M4 (AML – M4)
- CBFB/MYH11, inv(16)(p13;q22)

CHRONIC MYELOID LEUKEMIA (CML)
- BCR/ABL, t(9;22)(q34;q11)
- FGFR1, del(8p12)
- PDGFRβ, t(5;12)(q13;13)

HYPEREOSINOPHILIC SYNDROME
- FIPLI-PDGFRα, del(4q12)

MYELODYSPLASTIC SYNDROME (MDS)
- EGR1, del(5q31)
- del(7q31)
- Monosomy 7
- Trisomy 8
- D20S108, del(20q12)
- Nulisomy Y

MYELOPROLIFERATIVE DISORDERS (MPS)
- del(4q12)
- EGFR, del(5q31)
- Deletion 7q31 / Monosomy 7
- Monosomy 8p11.1-q11
- D20S108, del(20q12)
- Nulisomy Yp11.1
- BCR/ABL, t(9;22)(q34;q11)

MYELOPROLIFERATIVE NEOPLASMS
- JAK2 (exon 14) V617F mutation
- CALR
- MPL

THROMBOPHILIA
- Factor V (Leiden mutation)
- Factor II (2021A mutation)
- MTHFR (C667T mutation)
<table>
<thead>
<tr>
<th>Disorder</th>
<th>Genetic Alterations</th>
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<tbody>
<tr>
<td><strong>Acute lymphoblastic leukemia (ALL-B)</strong></td>
<td>MLL, del(11q23)</td>
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<tr>
<td></td>
<td>FLT3 (13q12.2)</td>
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<td></td>
<td>TCF3/PBX1, t(1;19)(q23;p13)</td>
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<td></td>
<td>BCR/ABL, t(9;22)(q34;q11)</td>
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<td></td>
<td>CMYC/IGH, t(8;14)(q24;q32)</td>
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<td>TEL/AML1, t(12;21)(p12;q22)</td>
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<td><strong>Chronic lymphocytic leukemia (CLL)</strong></td>
<td>ATM, del(11q22.3)</td>
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<td></td>
<td>MLL, del(11q23)</td>
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<td>Trisomy 12</td>
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<td>D13S25, del(13q14.3)</td>
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<td></td>
<td>RB1, del(13q14)</td>
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<td>TP53, del(17p13.1)</td>
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<td>IGH (14q32)</td>
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<td>BCL1/IGH, t(11;14)(q13;q32)</td>
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<td>IGH/BCL2, t(14;18)(q32;q21)</td>
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<td><strong>Multiple myeloma</strong></td>
<td>RB1, del(13q14)</td>
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<td>TP53, del(17p13.1)</td>
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<td>del(1p32)</td>
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<td>del(1q21)</td>
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<td>Trisomy 6</td>
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<td>Trisomy 9</td>
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<td>Trisomy 11</td>
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<td>Trisomy 15</td>
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<td>IGH, t(14q32)</td>
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<td>FGFR3/IGH, t(4;14)(p16;q32)</td>
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<td>BCL1/IGH, t(11;14)(q13;q32)</td>
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<td>IGH/MAF, t(14;16)(q32;q23)</td>
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<td><strong>Lymphoproliferative disorders (LPD)</strong></td>
<td>Clonal B-cell LPD (IGH)</td>
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<td>Clonal T-cell LPD (TCR)</td>
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<td><strong>Acute lymphoblastic leukemia (ALL-T)</strong></td>
<td>SIL/TAL1, t(1p32)</td>
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<td>TLX3, t(5q35)</td>
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<td>MYB, del(6q23)</td>
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<td>NOTCH1</td>
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<td><strong>MALT lymphoma</strong></td>
<td>BCL6, t(3;14)</td>
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<td>MALT1 (18q21)</td>
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<td>IGH/MALT1, t(14;18)(q32;q21)</td>
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<td>API2/MALT1, t(11;18)(q21;q21)</td>
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<td><strong>Diffuse large B-cell lymphoma (DLBCL)</strong></td>
<td>BCL6, t(3;14)</td>
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<td>CMYC (8q24)</td>
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<td>IGH (14q32)</td>
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<td>IGH/BCL2, t(14;18)(q32;q21)</td>
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<td><strong>Follicular lymphoma</strong></td>
<td>BCL6, t(3;14)</td>
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<td>CMYC (8q24)</td>
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<td>IGH/BCL2, t(14;18)(q32;q21)</td>
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<td><strong>Burkitt’s lymphoma</strong></td>
<td>CMYC (8q24)</td>
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<td>CMYC/IGH t(8;14)(q24;q32)</td>
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<td><strong>Mantle cell lymphoma</strong></td>
<td>CCND1/IGH, t(11;14)(q13;q32)</td>
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<tr>
<td><strong>Anaplastic large cell lymphoma (ALCL)</strong></td>
<td>ALK rearrangements (2p23)</td>
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</tbody>
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Please ask for more tests
- By FISH
- By Molecular Biology
### Solid Tumors (Epithelial-Estromal)

#### Esophagus
- HER2 (17q11.2-12)
- ZNF217 (20q13.2)
- CMYC (8q24)
- P16, del(9q21)

#### Ewing’s Sarcoma
- t(11;22)(q24;q12)
- t(21;22)(q22;q12)
- t(7;22)(p22;q12)
- t(2;22)(q33;q12)
- t(17;22)(q12;q12)

#### Synovial Sarcoma
- t(X;18)(p11;q11)

#### Clear-Cell Sarcoma
- t(12;22)(q22;q13)

#### Myxoid Chondrosarcoma
- BCR/ABL1, t(9;22)(q34;q11)

#### Myxoid Liposarcoma
- t(12;22)(q13;q12)

#### Dermatofibrosarcoma
- t(17;22)(q13;q12)

#### Gastrointestinal Stromal Tumor (GIST)
- PDGFRα

#### Desmoplastic Small Round Cell Tumor
- t(22q12)

#### Colorectal Cancer
- KRAS *
- NRAS *
- BRAF *
- PIK3CA *
- TP53 *
- EGFR
- APC
  * Performed by NGS
- TP53, del(17p13.1)
- del(17p11.2)
- BCL2, del(18q21)
- D7S486 (7q31)
- LPL, del(8p22)
- CMYC (8q24)
- ZNF217 (20q13.2)
- BCR, del(22q11)
### LUNG CANCER
- ALK, t(2p23)
- ROS1 (6q22)
- RET (10q11)
- MET (7q31)
- EGFR (7p12)
- TP53, del(17p13.1)
- EGFR
- KRAS

### BREAST CANCER
- HER2 (17q11.2-12)
- Topo IIα (17q21)
- ZNF217 (20q13.2)
- PTEN, del(10q23)
- MYC (8q24.2)
- EGFR (7p12)
- BRCA1 *
- BRCA2 *
- PIK3CA *
  * Performed by NGS

### NEUROBLASTOMA
- NMYC (2p24.1)
- MLL, del(11q23)
- 1p gain
- 11q loss
- 17q loss
- 3p loss

### BLADDER CANCER
- P16, del(9q21)
- Aneuploidy 3p
- Aneuploidy 7p
- Aneuploidy 17
- TP53, del(17p13.1)

### PROSTATE CANCER
- LPL, del(8p22)
- CMYC (8q24)
- TP53, del(17p13.1)
- PTEN, del(10q23)
- Chromosome X

### MELANOMA
- MYB (6q23)
- BCL1 (11q13)
- RREB1 (6p25)
- BRAF V600

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CONGENITAL DISORDERS & INHERITED DISEASES

HEREDITARY BREAST-OVARIAN CANCER
- BRCA1 *
- BRCA2 *
  * Performed by NGS

HEREDITARY NONPOLYPOSIS COLON CANCER (LYNCH SYNDROME)
- MLH1 *
- MSH2 *
- MSH6 *
- PMS2 *
  * Performed by NGS

HEREDITARY PANCREATIC CANCER
- ATM *
- BRCA2 *
- PALB2 *
  * Performed by NGS

RETINOBLASTOMA
- RB1

SPASTIC PARAPLEgia
- ATL1
- SPAST
- SPG31
- CYP7B1

HEREDITARY POLYPOSIS COLON CANCER
- APC *
- MUTYH *
  * Performed by NGS

HEREDITARY PROSTATE CANCER
- BRCA2 *
- CHEK2 *
- HOXB13 *
  * Performed by NGS

COWDEN SYNDROME
- PTEN *
- SDHB *
- SDHD *
- PIK3CA
  * Performed by NGS

FAMILIAL MALIGNANT MELANOMA
- CDKN2A

NEUROFIBROMATOSIS
- NF1
- NF2
**Duchenne Syndrome**
- DMD

**Hereditary Periodic Fever Syndromes**
- MEFV
- TRAPS

**Muscular Dystrophy / Spinal Muscular Atrophy**
- AR (expansion CAG)
- FSHD
- DMD

**Hypothyroidism**
- THRB

**Acidemia / Aciduria Methylmalonic**
- MMACHC

**Hereditary Fructose Intolerance (Aldolase B Deficiency)**
- MMACHC

**Fragile X Syndrome**
- FMR1 (triplet CGG region expansion)

**Familial Hypocalcaemia**
- CASR

**Cystic Fibrosis**
- CTFR

**Serpinopathies (α-1 Antitrypsin Deficiency)**
- CYP21A2

**Diabetes**
- AVPR2
- GCK
- HNF-1α

**Hereditary Pancreatitis**
- SPINK1
- PRSS1

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CONGENITAL DISORDERS & INHERITED DISEASES

DIGEORGE SYNDROME
- Tuple1, microdeletion del(22q11.2)

PRADER-WILLI AND ANGELMAN SYNDROMES
- D15S11, del(15q11)
- GABRB3, del(15q12)

MILLER-DIEKER SYNDROME
- L1S1, del(17p13)

MENTHAL RETARDATION
- 60K array CGH

HIGH RESOLUTION ANALYSIS
- 400K array CGH

ALZHEIMER
- APOE
- APP
- PSEN1
- PSEN2

CHARCOT-MARIE-TOOTH DISEASE
- MFN2
- PMP22

WILLIAMS SYNDROME
- Monosomy 7, del(7q11.23)

KALLMAN SYNDROME (HYPOGONADISM)
- KAL, del(Xp22.3)

CHROMOSOMES X AND Y ABNORMALITIES
- XIST, del(Xq13.2)
- SRY, del(Yp11.3)

AUTISM
- 180K array CGH

EXOME PROFILING
- Exome 100X
- Exome 50X

EPILEPSY
- PCDH1
- SCN1A
- Panel 122 genes*
  * Performed by NGS

HUNTINGTON DISEASE
- IT15

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INFECTIONOUS DISEASES

MONONUCLEOSIS / LYMPHOMA
- Epstein-Barr virus
- Citomegalovirus

WHOOPING COUGH / PERTUSSIS-LIKE SYNDROME
- Bordetella pertussis
- Bordetella parapertussis

WHIPPLE’S DISEASE
- Tropheryma whippelii
- Tropheryma whippelii

GASTRITIS / GASTRIC ULCER / GASTRIC CANCER
- Helicobacter pylori

HUMAN PAPILOMAVIUS
- Papilomavirus

HERPES, HERPES ZOSTER
- Herpes virus I y II
- Herpes virus VI
- Herpes virus VIII
- Varicela-zóster

LEPROSY / HANSEN’S DISEASE
- Mycobacterium leprae
- Mycobacterium spp.

TUBERCULOSIS
- Mycobacterium tuberculosis
- Mycobacterium spp.

CHAGAS DISEASE / AMERICAN TRYPANOSOMIASIS
- Trypanosoma cruzi

INFECTION ENDOCARDITIS (HACEK)
- Haemophilus parainfluenzae
- Haemophilus aphrophilus
- Actinobacillus actinomycetemcomitans
- Cardiobacterium hominis
- Eikenella corrodens
- Kingella kingae

POLIOMYELITIS, MENINGITIS (ENTEROVIRUS) S
- Poliovirus
- Coxsackie A
- Coxsackie B
- Echovirus
- Enterovirus 68-71

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