**Custom Genomic Analysis Form**

1. **CUSTOMER INFORMATION**

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| **Institución:** | **Fecha de envío:** |
| **Name:** | **Email:** |
| **Address:** |  |
| **Phone number:** |  |

1. **SAMPLE INFORMATION**

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|  | **SAMPLE TYPE (mark with an X)** | | | |
| **Sample Name** | **Annotated Variants (xcl)** | **Fastq File** | **DNA** | **Blood** |
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1. **REQUEST FOR ANALYSIS**

**1) Genetic analysis from annotated variants of Primary Immunodeficiency (PID) Sequencing Panel**

An analysis of the responsible pathology candidate variants is performed based on the patient's anonymous clinical data. A report is provided with the variants, their allelic frequency and the deleteriousness prediction based on the MSC.

The analysis time is one week

Price: 50 eur/sample

**2) Genetic analysis from annotated whole exome sequencing (WES) variants**

An analysis of the candidate gene variants responsible for the pathology is performed based on the patient's anonymous clinical data. A report is provided with the variants, their allelic frequency and the deleteriousness prediction based on the MSC.

The analysis time is a month and a half.

Price: 600 eur/sample

**3) Genetic analysis from annotated genome mass sequencing (WGS) variants**

An analysis of the candidate gene variants responsible for the pathology is performed based on the patient's anonymous clinical data. A report is provided with the variants, their allelic frequency and the deleteriousness prediction based on the MSC.

The analysis time is three months

Price: 1700 eur/simple

**4) Genetic analysis from raw data in FASQ format from PID Sequencing Panel**

Annotation of the variants and analysis of the candidate variants responsible for the pathology is carried out based on the patient's anonymous clinical data. A report with the variants, their allelic frequency and the prediction of deleteriousness based on the MSC is delivered.

The analysis time is one week

Price: 180 eur/sample

**5) Genetic analysis from raw data in whole exome sequencing (WES) FASQ format**

Annotation of the variants and analysis of the variants of the candidate genes responsible for the pathology is carried out based on the patient's anonymous clinical data. A report with the variants, their allelic frequency and the prediction of deleteriousness based on the MSC is delivered.

The analysis time is two and a half months

Price: 800 eur/sample

**6) Validation by Sanger sequencing of the selected variants**

At the user's request, the desired candidate variants can be validated by Sanger sequencing. To do this, the user will provide the platform with 1 microgram of DNA or 3mL of blood in EDTA. A report will be sent with the chromatographic image of the variant. The price includes the primers and all the reagents for the PCR. And in case the starting sample is blood, the reagents for DNA extraction and counting are included.

The analysis time is three months

Price: 180 eur/sample from DNA

Price: 220 eur/sample from blood in EDTA

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| **TYPE ANALYSIS** | **DURATION** | **PRICE PER SAMPLE** | **SELECTED (mark with X)** |
| **Genetic analysis from annotated variants of PID Sequencing Panel** | 1 week | 50 eur |  |
| **Genetic analysis from annotated whole exome sequencing (WES) variants** | 1.5 month | 600 eur |  |
| **Genetic analysis from annotated whole genome sequencing (WGS) variants** | 3 months | 1700 eur |  |
| **Genetic analysis from raw data in FASQ format from PID Sequencing Panel** | 1 week | 180 eur |  |
| **Genetic analysis from raw data in whole exome sequencing (WES) FASQ format** | 2.5 months | 800 eur |  |
| **Validation by Sanger sequencing of the selected variants** | 3 months | 180 eur/sample from DNA |  |
| 220 eur/sample from blood in EDTA |  |

1. **INSTRUCTIONS FOR SENDING SAMPLES**

The computer files will be sent to the email address rebeca.perez@idipaz.es

Large FASTQ files will be sent on hard disk or USB memory stick and delivered to the postal address:

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| Rebeca Pérez de Diego, PhD  Laboratory of Immunogenetics of Human Diseases  Office 3.2- 3rd floor- IdiPAZ Institute for Health Research  Hospital La Paz. Pº Castellana, 261, 28046, Madrid, Spain | Rebeca Pérez de Diego, PhD  Laboratorio de Inmunogenética de las Enfermedades  Despacho 3.2, 3ª planta Instituto de Investigación Sanitaria Hospital La Paz-IdiPAZ  Hospital La Paz, Pº Castellana, 261, 28046, Madrid, España |

DNA or blood samples will be sent to the address:

|  |  |
| --- | --- |
| Rebeca Pérez de Diego, PhD  Laboratory of Immunogenetics of Human Diseases  Office 3.2- 3rd floor- IdiPAZ Institute for Health Research  Hospital La Paz. Pº Castellana, 261, 28046, Madrid, Spain | Rebeca Pérez de Diego, PhD  Laboratorio de Inmunogenética de las Enfermedades  Despacho 3.2, 3ª planta Instituto de Investigación Sanitaria Hospital La Paz-IdiPAZ  Hospital La Paz, Pº Castellana, 261, 28046, Madrid, España |