BIOARRAY
Diagnóstico Genético

Informed Consent for Genetic Testing

Genetic analysis requested:	
Clinical Indication / Diagnostic Suspicion:	

1. Description, purpose and benefits of the analysis

A genetic analysis consists of the study of the patient's genetic material (DNA, RNA or chromosomes) in order to identify variants that may explain a clinical picture, confirm a diagnosis, assess the risk of developing a disease, guide medical management or facilitate family genetic counseling. The specific technique used (genomic microarray, Sanger sequencing, MLPA, next-generation sequencing gene panel, exome or genome, expansion, methylation, karyotype, FISH or other studies) is determined by the clinical indication and is detailed in the section "Genetic analysis requested" and in the test requisition form.
The results may be positive (explanatory variant of the phenotype, or which increases the risk of disease), negative (no relevant variants detected) or uncertain (variants of unestablished clinical significance). A negative result does not absolutely rule out a genetic cause; a positive result does not always guarantee direct clinical utility. The results should always be interpreted by a physician in the patient's clinical context.
Where indicated by the prescribing physician in the requisition form, the scope of the analysis may be extended internally without requiring a new sample or new signature from the patient (e.g., transition from virtual panel to whole exome in extensive sequencing-based analyses). This consent covers these extensions when they are provided for in the requisition.

2. Procedure

2.1 How is the analysis performed?

A biological sample of the patient is required—usually peripheral blood in a tube with EDTA-K, Li-Heparin, and, depending on the test, also saliva, buccal swab, biopsy, or other tissues—from which genetic material is obtained. Blood collection carries only the minimal risks of venipuncture and does not pose an added risk to health. In the laboratory, the sample is processed using the appropriate technique as indicated by the prescribing physician; Clinically relevant results are confirmed, where appropriate, by orthogonal technique.

2.2 Which relatives are analyzed?

Depending on the family structure and clinical suspicion, it may be necessary to test one or more biological relatives to establish the origin and significance of the variants. If, during the analysis, incompatibility is detected between the declared family ties and the biological relationships observed (non-paternity, false sibling relationship, etc.), this finding will not be reported in the report, unless it is essential for the correct interpretation; in this case, it will be transferred to the prescribing doctor with the utmost confidentiality.

2.3 Genetic counseling and delivery time

In accordance with Article 54 of Law 14/2007 on Biomedical Research (LIB), all genetic analysis for health purposes must be accompanied by prior and subsequent genetic counselling, by the prescribing physician or, where appropriate, by professionals with specific training in clinical genetics. The turnaround time of the report depends on the technique used and is communicated when the analysis is requested. This period may be affected by pre-analytical incidents, complexity of the case, additional studies in relatives or possible extensions of the analysis.

3. Limitations of the study

3.1 Range and sensitivity

No genetic technique detects 100% of possible variants. The scope of the analysis is limited to the genes, regions or types of variant covered by the technique requested by the prescribing physician. Certain types of variants (copy number variants, structural rearrangements, mosaicisms below analytical sensitivity, repeat expansions, variants in repetitive regions, epigenetic modifications, uniparental disomies, variants in deep regulatory or intronic regions) may not be detected or may be detected with reduced sensitivity, depending on the technique used. When clinical suspicion so advises, the prescribing physician may request complementary techniques.

3.2 Erroneous results

Analytical sensitivity and specificity are less than 100%. Exceptionally, false positives or false negatives may occur. If the cause of a discordant result cannot be attributed to a traceable laboratory error, Bioarray will not be responsible for the incomplete or equivocal result.

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4. Results report

4.1 Interpretation and content of the report

Variants are classified according to the international guidelines applicable to each variant type and technique (ACMG, ClinGen, ISCN, etc., as appropriate). The report shall include, at a minimum: identification of the patient and the sample, clinical indication, technique used and scope of analysis, applicable quality metrics, pathogenic and likely pathogenic variants related to the phenotype, variants of uncertain significance when clinically relevant, segregation when studied, limitations of the analysis and clinical recommendations.

4.2 Default Findings

By default, the report will only include variants that can explain the phenotype within the requested scope. Findings in non-indicated genes, pharmacogenetic variants or unrelated carrier status will not be reported, unless expressly authorised by the patient in section 9 (Optional consents).

4.3 Report recipients and raw data

The report is delivered to the prescribing physician. Upon request, a copy may be sent to other healthcare professionals involved in the patient's clinical care or to the requesting laboratory. When the technique generates it, the prescribing physician may request the raw data of the analysis, which may contain more information than that analyzed and reported by Bioarray; the responsibility for their subsequent interpretation lies with the person requesting them.

5. Reanalysis and updating of results

Knowledge about genetic variants evolves over time. As part of Bioarray's commitment to service quality, the laboratory will be able to review previous cases when new scientific evidence may substantially modify a result already issued. This review is part of the contracted service and does not require additional authorisation. Where appropriate, Bioarray will issue an updated report to the prescribing physician, unless the patient, in exercise of his or her right not to know (art. 4.5 LIB), has expressed in writing his or her wish not to receive this supervening information. The patient or prescribing physician may also actively request a reanalysis, recommending waiting at least twelve months from the original analysis or at any time if the clinical phenotype has changed; This request may involve an additional cost, previously communicated.

6. Study on minors

The following safeguards apply to minor patients (Law 14/2007 and ESHG/ACMG recommendations): consent will be granted by both parents or legal representatives, except in the case of accredited single-parent families or sole guardianships; minors < 16 years of age or unemancipated shall consent by proxy, with a hearing of the minor according to his maturity; Mature minors (≥ 16 years of age or emancipated) will consent on their own, in addition to parental consent. In asymptomatic minors, variants associated with adult-onset diseases without effective preventive or therapeutic measures will not be reported, preserving the minor's right to decide when they reach the age of majority.

7. Data protection, storage and use in research

Your personal, health and genetic data are special category data (art. 9 GDPR) and will be treated with strict confidentiality by BIOARRAY S.L. in accordance with the GDPR, the LOPDGDD 3/2018, Law 14/2007 (LIB) and Law 41/2002. Access is restricted to authorised personnel and, where required by law, to the competent authorities (art. 49 LIB). Bioarray will not communicate your data to third parties, except for legal obligation or necessary subcontracting of qualified collaborating laboratories within the European Economic Area, under a confidentiality agreement.

- Legal bases: explicit consent (art. 9.2.a GDPR), execution of the service contract (art. 6.1.b GDPR), provision of healthcare by a professional subject to secrecy (art. 9.2.h GDPR) and, where appropriate, scientific research (art. 9.2.j GDPR and DA 17 LOPDGDD). Purposes: carrying out the analysis, quality control and assurance, communication with the prescribing doctor, compliance with legal obligations and, if authorised, research.
- Retention: the data will be kept for at least five years from the end of the analysis (art. 50 LIB), extendable in accordance with the health regulations on medical records (art. 17 Law 41/2002) and the deadlines required for accreditation and quality. Samples will be retained for the time strictly necessary for service or, if authorized, for research.
- Rights: you can exercise your rights of access, rectification, deletion, opposition, limitation and portability, as well as withdraw your consent, at any time by writing to BIOARRAY S.L. (Parque Científico y Empresarial de la UMH, Edificio Quorum III, S/N, 03202 Elche, Alicante; info@bioarray.es). Data Protection Officer: Andrés Antón Salas. In accordance with Article 60 LIB, you can revoke consent and request the irreversible destruction or dissociation of your samples, unless they have already been irreversibly anonymised; The revocation does not affect the lawfulness of the previous processing or the results issued. Complaint to the Spanish Data Protection Agency (www.aepd.es).

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8. Patient Statement

I DECLARE THAT, HAVING READ AND UNDERSTOOD THE ABOVE, I AM INFORMED:
I have received sufficient information about the nature, purpose, procedure, scope, limitations and possible results of the requested analysis, as well as about the processing of my data. I have had the opportunity to clarify my doubts and they have been resolved to my satisfaction.
Consequently, I GIVE MY FREE, VOLUNTARY AND INFORMED CONSENT to: (1) the performance of the genetic analysis indicated in this document by the prescribing physician and, if applicable, its internal extension in accordance with the request; (2) the processing of my personal, health and genetic data by BIOARRAY S.L. for the purposes of section 7; (3) the communication of the results to the prescribing physician and, at his request, to other professionals involved in my clinical care; and (4) the possible subcontracting of specific phases of the analysis to qualified collaborating laboratories within the EEA, with Bioarray maintaining responsibility for the final result.
This consent may be revoked at any time in writing, without affecting the lawfulness of the previous processing or the results already issued. Until such time as I am informed of the results, I have the right not to be informed of them (right not to know, art. 4.5 LIB) and to request the destruction of the sample (art. 60 LIB); I can also express in writing, through my prescribing doctor, my desire not to receive information arising from future re-analyses.

9. Optional Consents

The following consents are VOLUNTARY and INDEPENDENT of each other. Its refusal does not affect the performance of the analysis or the quality of the report. By default, the lab will NOT report the categories described below; only those for which the patient or their legal representative expressly marks "Yes" will be included. Not checking any boxes is considered "No." Some categories are only applicable when the technique used allows analysis (e.g., ACMG SF secondary findings or pharmacogenetic variants require extensive sequencing techniques); The laboratory will apply only the options consistent with the technique performed.

9.1 Secondary findings — ACMG SF list

List of clinically actionable genes from the American College of Medical Genetics and Genomics (ACMG), in which there are effective prevention, screening or treatment measures when identifying a pathogenic or likely pathogenic variant. Applicable in exome or genome-based analyses. The current version (currently ACMG SF v3.3 or later, www.acmg.net) will apply.

☐ **Yes** ☐ **No** I CONSENT to be informed of the secondary findings identified in the genes of the current ACMG SF list.

9.2 Incidental findings outside the ACMG SF list

Pathogenic variants detected in an unsought manner in genes outside the requested scope and the ACMG SF list, but with significant clinical implication for the patient or their relatives. The finding will be evaluated by the Bioarray Interpretation Committee and, if the patient authorizes it, communicated to the prescribing physician.

☐ **Yes** ☐ **No** I AUTHORIZE the communication to the prescribing physician of incidental findings with significant clinical relevance outside the requested scope and the ACMG SF list.

9.3 Adult-onset findings with no clinical action available (adults only)

Variants associated with adult-onset diseases for which there is currently no effective preventive or therapeutic intervention (e.g., genetic forms of late-onset Alzheimer's disease). Category available only for adults and in techniques that allow its analysis.

☐ **Yes** ☐ **No** I CONSENT to be informed of adult-onset findings with no clinical action available.

9.4 Carrier status of recessive diseases

A recessive disease (e.g., cystic fibrosis) requires pathogenic variants in both alleles of the gene to manifest. A person carrying a single variant does not have the disease, but can transmit it to their offspring. If a variant also explains the patient's phenotype, it will be reported regardless of this option.

☐ **Yes** ☐ **No** I CONSENT to be informed of my status as a carrier of pathogenic variants in genes associated with recessive diseases.

9.5 Pharmacogenetic variants

Variants that do not directly produce pathology, but can influence the body's response to certain drugs. Applicable in exome or genome-based analyses; it should be interpreted in conjunction with the responsible physician.

☐ **Yes** ☐ **No** I CONSENT to be informed of the pharmacogenetic variants detected during the analysis.

9.6 Sample conservation and use for research

Authorisation of the storage of an aliquot after the completion of the analysis and its pseudonymised or anonymised use in research projects, validation of techniques and improvement of the service. It does not entail economic benefit for the patient and can be revoked at any time (art. 60 LIB).

☐ **Yes** ☐ **No** I CONSENT to the conservation and use of my sample and data in research, pseudonymized or anonymized. Otherwise, the sample will be destroyed at the end of the analysis, except for legal storage periods.

10. Confirmation of the prescribing physician

The prescribing physician acknowledges that: (1) he has provided the patient or his legal representative with the relevant clinical information about the requested analysis and its scope, including the genetic counseling required by art. 54 LIB; (2) has obtained informed consent from the patient or legal representative, by signing this document or kept in the medical record; (3) the patient has not exercised the right not to know to date; and (4) promptly transfer to Bioarray any subsequent request for revocation or exercise of rights.

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11. Signatures

Having read and understood the information provided, I sign this document. Please sign two copies: return one to the lab along with the sample and keep the other for your records.

Patient / Legal Representative	**Prescribing physician**
Name and surname:	Name and surname:
ID number:	Medical registration number:
Signature and date:	Signature and date:

In the case of a minor patient or with judicially modified capacity, signature of both parents or legal representatives:

Parent / Representative 1	**Parent / Representative 2**
Name and surname:	Name and surname:
ID number and kinship:	ID number and kinship:
Signature and date:	Signature and date:

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