Limitations of Molecular Testing Consent Form

GC:
We, the undersigned, request prenatal diagnosis (PND) / preimplantation genetic diagnosis (PGD) for the variant identified in the gene ..... in our child ..... / in myself ..... / in my spouse ..... according to the American College of Medical Genetics and Genomics (ACMG) standards, this variant is classified as:

☐ **Likely Pathogenic:** This variant is classified as likely pathogenic, meaning it is approximately 90% likely to be disease-causing.
☐ **Variant of Uncertain Significance (VUS):** This variant is classified as VUS, meaning its clinical significance is unclear. Therefore, its pathogenicity has not been definitively established.
Hence, the causality of this variant is not yet confirmed and prenatal diagnosis based on this variant has limitations.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ No clinical examination was performed in this center before the death of our child, and no medical records confirming the disease in our child exist; therefore, clinical correlation was not possible.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ Clinical correlation between the identified gene variant and disease was not performed before the death of our child; thus, clinical correlation was not possible.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ Since definitive clinical correlation between the identified variant and our child's symptoms was not possible, if our child's problems are related to this gene or other genes, prenatal diagnosis based on this variant may not be informative.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ The pathogenic gene variant was identified by another center, and we are only verifying the identified variant; we cannot comment on the accuracy or completeness of the initial testing.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ The decision for therapeutic abortion based on the identified variant rests with the legal medical authorities, and this center bears no responsibility for abortion decisions.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ Since the variant ..... has not previously been confirmed by Sanger sequencing in this center, during primer design we may find that confirmation is not feasible, in which case mutation confirmation testing of the couple and fetus will be canceled and mutation confirmation fees, except DNA extraction, will be refunded.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ Because the variant ..... has not been previously confirmed by Sanger sequencing at this center, primer design and ordering may prolong test turnaround time, possibly exceeding the legal abortion timeframe.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ Since amniotic fluid was collected for molecular testing, DNA extraction and fetal genetic analysis may take enough time to miss the legal abortion window; therefore, testing is performed primarily for reassurance.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ In approximately 1% of chorionic villus sampling (CVS) cases, maternal cell contamination (MCC) may occur. MCC testing can verify if the sampled cells belong to the fetus or mother. If maternal contamination is detected, repeat sampling is recommended.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ In cases of bloody amniotic fluid samples, maternal cell contamination may be present and detectable by MCC testing. Bloody samples may also result in longer test times or require repeat sampling.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

☐ In twin pregnancies, twin-QF PCR testing is recommended before molecular testing. Identical DNA profiles may reflect monozygotic twins or sampling from only one sac; the sampling physician will decide if repeat sampling is necessary.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

It has been explained to me/us that the technical results of the performed test may be used for scientific and research purposes, provided that all identifying personal information remains confidential. I/we hereby give my/our consent for such use.

It has also been clarified that the laboratory is obligated to provide technical information to relevant national authorities upon request, in accordance with the regulations approved by the Ministry of Health.

I/we, the undersigned, after reading all the items stated in this consent form, receiving the necessary explanations, and having sufficient opportunity to ask questions, hereby knowingly and voluntarily—without any pressure or coercion—and with full understanding of the possible outcomes of the mentioned tests and their potential emotional, psychological, and physical consequences, sign this consent form and request the center to proceed with the testing.

I understand that once the test has been accepted, it cannot be cancelled.

☐ I hereby give my consent for the test to be performed.

Date: ..... / ..... / ..... Signature (Female): _____ Signature (Male): _____

Date: ..... / ..... / ..... Name and Signature of Genetic Counselor: _____

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