

# PGD Patient Referral Form



Please submit form by EMAIL: [ivf@combimatrix.com](mailto:ivf@combimatrix.com) or FAX:949.753.4725

## Clinic Information

Clinic Name \_\_\_\_\_ Referring Physician \_\_\_\_\_  
Address \_\_\_\_\_ Physician Signature \_\_\_\_\_  
City \_\_\_\_\_ State \_\_\_\_\_ ZIP \_\_\_\_\_ Clinical Contact \_\_\_\_\_  
Tel \_\_\_\_\_ Fax \_\_\_\_\_ Email \_\_\_\_\_

## Patient Information

Patient's Last Name \_\_\_\_\_ First Name \_\_\_\_\_ Date of birth \_\_\_\_/\_\_\_\_/\_\_\_\_ Gender:  F  M  
Partner's Last Name \_\_\_\_\_ First Name \_\_\_\_\_ Date of birth \_\_\_\_/\_\_\_\_/\_\_\_\_ Gender:  F  M  
Address \_\_\_\_\_ City \_\_\_\_\_ State \_\_\_\_\_ ZIP \_\_\_\_\_  
Tel \_\_\_\_\_ Cell \_\_\_\_\_ Email \_\_\_\_\_  
Egg donor used?  No  Yes *If yes, donor age* \_\_\_\_\_ Male factor?  No  Yes *If yes:*  TESE  ICSI  Donor sperm

## Single Gene Disorders *(For chromosome translocations, complete the section below this one)*

» ENCLOSE REPORTS «

Disorder Name \_\_\_\_\_ **MATERNAL Status:**  Affected  Symptomatic  Asymptomatic  
 Carrier  Affected  Unaffected  
 Non-Carrier  
Affected/Tested Family Members \_\_\_\_\_  
\_\_\_\_\_ **PATERNAL Status:**  Affected  Symptomatic  Asymptomatic  
\_\_\_\_\_  Carrier  Affected  Unaffected  
\_\_\_\_\_  Non-Carrier

## Chromosome Translocations *(For single gene disorders, complete the previous section)*

» ENCLOSE REPORTS «

**MATERNAL Status:**  Normal karyotype  Balanced chromosome translocation **PATERNAL Status:**  Normal karyotype  Balanced chromosome translocation  
Karyotype \_\_\_\_\_ Karyotype \_\_\_\_\_

## TESTING PROCESS OVERVIEW

**STEP 1 PATIENT REFERRAL** Email or fax a completed *PGD Patient Referral Form* along with documentation of the familial disorder (i.e. molecular test results indicating the pathogenic mutation for single gene disorders or cytogenetics reports showing the chromosomal abnormality for chromosome translocations). Please obtain these records prior to referring your patient to prevent delays in the testing process.

**STEP 2 INITIAL GENETIC COUNSELING** Once your referral has been received by CombiMatrix, one of our board-certified Genetic Counselors will schedule an initial telephone consultation with your patient and her partner to obtain all necessary information and test results prior to the Medical Team review.

**STEP 3 MEDICAL TEAM REVIEW** *Single Gene Disorders* – Following the initial genetic counseling consultation, the patient's case will be reviewed by our Medical Team based on the specific gene, familial mutation(s), family history, and whether one or more appropriate relatives are available to provide a reference DNA sample. *Chromosome Translocations* – Following the initial genetic counseling consultation, the patient's case will be reviewed by our Medical Team based on the chromosomal abnormality and family history to determine whether the patient is a candidate for PGD.

**STEP 4 INFORMED CONSENT** Once the patient's case has been approved, one of our board-certified Genetic Counselors will schedule a follow-up phone consultation to discuss patient-specific risks and review the benefits, limitations and risks of PGD. For single gene disorders, DNA collection kits will be mailed to the patient, partner and any reference family members required.

**STEP 5 VALIDATION OF TEST PERFORMANCE** (NOTE: *For single gene disorders only*) Once the patient's, partner's, and any reference family members' DNA samples have been received, they will be run for validation of the test's performance. If it is determined that a different reference sample is required for optimal test performance, the patient will be notified immediately. Once validation is complete, your office and your patient will be notified that CombiMatrix is ready to accept embryo biopsy samples.

**STEP 6 EMBRYO BIOPSY ANALYSIS** Following the receipt of the embryo biopsy samples, testing begins immediately, with results available in 10 days. A post-result telephone consultation can be arranged with one of our board-certified genetic counselors if your patient so desires.