

Preparing for Your First Reproductive Genetic Counseling Session

Patient Checklist

1. Clarify Your Goals & Narrative

Before your session, take a moment to define what you want to achieve. Having a clear focus helps your genetic counselor tailor the conversation to your specific emotional and medical needs.

- ☐ **Draft a "One-Line Goal":** Write down a single, clear sentence summarizing what you hope to get out of the appointment (e.g., *"I want to understand if we need extra embryo testing because of my family history,"* or *"I want to understand what our high-risk screening result means for this pregnancy"*).
- ☐ **Summarize Your Journey:** Briefly write down the key events that led you to schedule this appointment. This could include:
 - Fertility challenges or the length of time you have been trying to conceive.
 - A referral from your IVF or OB team due to age, ultrasound findings, or blood work.
 - A history of pregnancy loss or recurrent miscarriages.
- ☐ **Identify Your Boundaries:** Reflect on your personal feelings regarding testing. Remember that preconception and prenatal testing are entirely optional. Think about:
 - How having more genetic information might affect your anxiety or peace of mind.
 - Whether certain genetic results would change your family-building decisions or pregnancy management plans.

2. Gather Essential Medical & Genetic Records

Having your physical test results is one of the most critical parts of prep. Even if two people test as carriers for the same condition, genetic counselors look at the highly specific, technical variants to determine your actual clinical risk.

- ☐ **Compile Official Genetic Test Reports (Crucial):** Obtain copies of any genetic tests you or your partner have already completed. Do not rely on a doctor's verbal summary. Ensure you have the actual lab sheets for:
 - Carrier screening reports (e.g., screening for conditions like cystic fibrosis or sickle cell disease).
 - Pre-implantation Genetic Testing (PGT) reports if you have already undergone IVF (including PGT-A for chromosome numbers or PGT-SR for structural rearrangements).
 - Prenatal screening or diagnostic results (e.g., cell-free DNA blood work, amniocentesis reports, or CVS results).
- ☐ **Summarize Your IVF/Fertility History:** If you are working with an IVF clinic, have a high-level summary of your treatment history ready. You do not need highly technical cycle logs (like daily follicle measurements), but your counselor will want to know:
 - How many IVF stimulation cycles you have completed.
 - The number of embryos created and any previous testing outcomes.
- ☐ **Obtain Relevant Pregnancy Records:** If you have experienced pregnancy loss, bring any medical notes or pathology reports from those pregnancies. While over 50% of early losses are chromosomal, understanding your history helps your counselor discuss the clinical gaps and potential biological or non-biological factors.

3. Map Your Family History

A standard genetic counseling session involves a detailed, multi-generational family history. Collect these details from close relatives (parents, siblings, aunts, uncles, cousins, and nieces/nephews) before your appointment if possible.

- ☐ **List Family Diagnoses:** Note if anyone in your family has been diagnosed with:
 - Intellectual or learning differences.
 - Developmental delays.
 - Congenital physical features, such as heart defects or other anatomical anomalies.
- ☐ **Identify Known Genetic Conditions:** Find out if any blood relatives are known carriers of or have been diagnosed with inherited conditions (e.g., cystic fibrosis, spinal muscular atrophy, fragile X, or translocations/"funky chromosomes").
- ☐ **Note Family History of Reproductive Challenges:** Ask (if comfortable) if close relatives have experienced:
 - Recurrent pregnancy losses or stillbirths.
 - Significant fertility struggles or early menopause.

4. Ground Your Expectations

As you head into your meeting, keeping these clinical realities in mind can help you navigate the complex emotional landscape of genetic medicine:

- **Screening vs. Diagnostic Certainty:** Remember that tests like PGT-A or prenatal blood screenings are *screening tools*, not 100% guarantees. While they have excellent detection rates (often between 90% and 99%), they cannot eliminate all risks or fully guarantee a successful pregnancy.
- **The Power of "It Depends":** Genetics is highly variable, and every biological pathway is unique. Your counselor's role is to help you weigh the emotional, physical, and financial costs of your options, and empower you to make the choice that feels right for you.

5. Connect with Community Support

Navigating the genetic and emotional complexities of family building can feel isolating, but you do not have to walk this challenging journey alone. Connecting with others who share similar paths can provide profound comfort, healing, and guidance.

- **AllPaths Support Groups:** AllPaths Family Building offers specialized [peer groups](#) that can be incredibly valuable for navigating the coping process and balancing hope with realism during your journey. The IVF Group is a helpful space to connect with others for many who are doing IVF for any reason, including for genetic testing purposes.
- **Targeted Support for Rare Conditions:** If you are navigating an incredibly rare genetic diagnosis or variant, there are specialized support groups and podcasts dedicated to distinct, rare conditions [51]. The [Undiagnosed Diseases Network \(UDN\)](#) is another wonderful, dedicated network focused on rare diseases that can provide personalized guides and a huge network of patient and scientific support.