Intermountain Precision Genomics

Intermountain MFM Prenatal Genetic Counseling Key Messages

This document serves to outline genetic counseling services in Maternal Fetal Medicine and provide formalized instructions for the most updated processes. We hope it is useful as a reference for clinic managers, patient service representatives, and providers in daily decision making, training new hires, organizing and strategizing, in huddle meetings, or when any questions arise regarding genetic counseling. Many decisions are made on a case by case basis, so if in doubt, please don't hesitate to reach out and ask. Let us know if there is information in this document that need clarification or updating.

- Intermountain MFM Prenatal GC team
  Updated October 2022

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1. Indications for Genetic Counseling Consultations:

Abnormal, Atypical, or No Result cell-free DNA (cfDNA or NIPS) results: The ordering provider informs the patient of the high-risk result. A GC is available to speak with outside providers and/or patients right away to discuss the results and present options. An MFM consultation, ultrasound, and further testing may then be coordinated.

Abnormal Maternal serum screen/MSS (quad): The ordering provider informs the patient of the high-risk result. A GC is available to speak with the patient before or in tandem with an MFM consultation and ultrasound.

Multiple gestation: An appointment with a GC may be scheduled to discuss testing options in twin or high multiple pregnancies if requested by the MFM or obstetric provider.

Previous affected pregnancy: If a provider has determined that a genetic counseling appointment is desired and warranted, a GC video/telehealth appointment may be scheduled. A referral to an MFM clinic may be initiated after this appointment by the genetic counselor.

Family history/familial testing/partner positive carrier screen: If a provider has determined that a genetic counseling appointment is desired and warranted, a GC video/ telehealth appointment may be scheduled. An appointment with an MFM provider may be coordinated for continued care.

Abnormal ultrasound findings:

- Indications for MFM AND a GC consult at the same appointment
  - Fetal lethal or grave conditions
  - Complex ultrasound findings: For example, cystic hygroma, ambiguous genitalia, abnormal brain, limbs, face, heart, skeletal anomalies, etc.
- Indications for **JUST** an MFM: The MFM will add a GC on if warranted or desired
  - Soft markers seen on outside ultrasound: echogenic intracardiac foci (EIF) or echogenic bowel, UTD (also known as pylectasis), mild ventriculomegaly, choroid plexus cysts, short femurs
  - Alloimmunization and other incompatibility
  - CPAM or gastroschisis
  - Abnormal placenta, uterus, or umbilical cord
  - Fetal Demise
  - Factor V Leiden or other thrombophilia concern
  - Teratogen exposure

Preconception Counseling:

- MFMs may determine that GC services are appropriate during their consultation and will have the GC join at that time or at a separate appointment
- Recurrent pregnancy loss (RPL): GC only appointments with referral from provider or MFM
- Consanguinity: warranted if closer than second cousins
- A patient with a genetic condition or at risk for a genetic condition for pregnancy management or reproductive purposes

Pregnancy Termination:

- GCs assist in coordinating if clinical indications are warranted
- Required Department of Health forms for all terminations are sent to our office for proper submission and tracking

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2. Referrals and Appointments: Utilizing the iCentra Pool "Intermountain Maternal Fetal Medicine- Genetic Counselors"

- Referrals come to MFM by email, fax, iCentra message, and self-referrals.
- For each referral, message the iCentra pool with the indication and other relevant information.
- A GC will respond quickly with appointment type, location, timing, or providers to be involved in the case.
- Same day appointments may be scheduled for urgent cases. Please confirm availability and timing with the GC staff by calling directly or messaging through teams.
- Consultation appointments are often added on the same day to an existing MFM appointment (abnormal ultrasound findings, new diagnosis, family history, testing of FOB, etc). PSRs assist with creating these FINs and checking the patient in/out.
- All high-risk or abnormal genetic screen results should have an appointment with a GC, even if not specified on the referral.

3. Exclusion criteria for a GC only consultation:

We do not typically see the following types of indications, but we would be happy to provide information and resources.

- General genetics screening: (low risk pregnancies, advanced maternal age only (AMA), or
- screening after PGT)
- Carrier screening (unless both partners are carriers of the same condition
- Cancer related genetic screening
- Fetal demise
- Neonatal/pediatric cases
- Family history of autism (unless there is also a history of recurrent pregnancy loss, intellectual disability, or other anomalies)
- Ehlers-Danlos syndrome (unless there is an identified vascular variant, personal or family history of aneurysm, organ rupture, or significant cardiac involvement)
- Paternity
- Maternal conditions (hypertension, diabetes, exposures, Graves disease, Crohn's disease, etc.)
- Antigen titers or incompatibility
- *MTHFR* testing or discussion

4. Returns of Referrals:

Some referrals may be returned to the ordering provider or edited to for a perinatologist only due to the above-mentioned exclusion criteria. In line with our mission to provide support, education, tools, and recommendations to outside clinics and communities, in these circumstances our office provides a brief explanation of why the referral is returned, along with possible recommendations and/or options for support from the GC staff. Please use the following explanations when returning a referral:

General Screening, screening in low-risk pregnancies, screening for advanced maternal age, and screening after PGT:

- OB/GYN offices offer genetic screening, including informed consent, test coordination, and return of results. Please let our GC staff know if your clinic would benefit from a educational meeting to discuss current recommendations, up to date information on testing options, test ordering, or the testing process. (Screening for fetal chromosomal abnormalities: ACOG Practice Bulletin Summary, No. 226. *Obstet Gynecol*. 2020;136(4):859-867)

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Carrier screening:

- Individually inherited conditions are rare, but collectively affect 1 in 300 pregnancies. Medical societies recommend carrier screening be offered to every patient regardless of ancestry. Whether the minimum number of conditions (SMA, CF, Fragile X, and hemoglobinopathies) or a larger expanded carrier screen panel is offered, the choice is up to the discretion of the provider and requires consent from the patient. Please let our GC staff know if your clinic would benefit from an educational meeting to discuss current recommendations, up to date information on testing options, test ordering, or the testing process.
  (Committee Opinion No. 690. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2017;129:e35–40; Committee Opinion No. 691. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2017 Mar;129(3):e41-e55)
- Exceptions: Please refer patients for a GC appointment in the following circumstances:
  - Both partners are carriers of the SAME condition
  - Patients who carry an X-linked condition (if known)
  - Patients who had a previous pregnancy affected with a genetic condition and desire counseling related to a current or future pregnancy

Fetal Demise:

- The Society for Maternal Fetal Medicine (SMFM) recommends that microarray analysis on POC be offered to patients with a fetal demise. This can be ordered by the provider. Genetic counselors are available to discuss how to coordinate testing, lab options, results review, clinical utility, and cost/insurance coverage if desired.

Neonatal/pediatric cases:

- It is often out of the scope of practice as perinatal GCs to counsel neonatal or pediatric cases. Families are referred to a medical geneticist at the Intermountain Genetics Clinic, Primary Children's Hospital, or the general Genetics Clinic at the University of Utah.

Family history of autism (unless there is also a history of recurrent pregnancy loss, intellectual disability, or other anomalies):

- Autism is more frequently diagnosed in males and occurs in about 1.5% of all children. Common genetic causes of autism include Fragile X syndrome, Rett syndrome, and various chromosomal deletion syndromes. Research suggests that even non-syndromic autism has a hereditary component, and the recurrence risk is approximately 5% for siblings of a case that is not associated with a known genetic condition. This risk is reduced for more distant relatives. No prenatal genetic testing for non-syndromic autism is available at this time. A referral may be made to an autism specialist.

Ehlers-Danlos Syndrome (unless there is identified history of aneurysm, organ rupture, or significant cardiac involvement):

- Ehlers Danlos syndrome (EDS) is a heterogeneous group of connective tissue disorders characterized by hypermobility, skin extensibility, and tissue fragility. There are many types of EDS classified according to the clinical manifestations.

Paternity:

- Testing for paternity is not performed in our MFM offices. Patients are encouraged to seek testing through the DNA Diagnostic Center at DNAcenter.com. The genetic counselors have additional information about this option by request.

Maternal conditions (hypertension, diabetes, exposures, Graves disease, Crohne's disease, etc.):

- These conditions are best monitored and treated by the primary OB/GYN or perinatologist with the exception of identified maternal genetic conditions.

Antigen titers, alloimmunization, or other incompatibility:

- A GC may be asked to assist with non-invasive genetic testing or diagnostic procedures. Cell free DNA testing is available for fetal antigens C, c, D, E, Duffy (Fya), and Kell (K)

***MTHFR* testing or discussion:**

- Recent publications have indicated that *MTHFR* polymorphisms are commonly observed in the general population and not causative for disease. Several medical societies such as the American College of Medical Genetics (ACMG), American Heart Association (AHA), and American College of Obstetrics and Gynecology (ACOG) have issued professional guidelines discouraging testing for these *MTHFR* polymorphisms. (ACOG Practice Bulletin No. 197: Inherited Thrombophilias in Pregnancy, Obstetrics & Gynecology: July 2018 - Volume 132 - Issue 1 - p e18-e34; Greenland P, et al. (2010) American College of Cardiology Foundation/ American Heart Association Task Force on Practice Guidelines. 2010 ACCF/AHA guideline for assessment of cardiovascular risk on asymptomatic adults. Circulation;*122*:e584–e636)

5. Testing kits:

Each clinic is supplied with frequently ordered testing kits. Site specific information should be discussed between the clinic manager and the GC staff to determine who should monitor when kits expire, when stocks are getting low, how new kits are ordered, and other logistics.

6. Test results in the medical record:

If possible, please separate out genetic test results into documentation as type "Genetics Lab Report" when they are received by way of a referral, outside prenatal records, or fax unless they are already in the medical record.

7. Communicating with GCs:

- Message through the medical record if the content should be documented in the patient's chart
- Group email: prenatalgc@imail.org Please use this for questions, referrals, or faxes that need to go to any member of our GC group.
- Group fax: **801-442-0592**
- Teams messaging: Please use teams messaging as an informal way of communicating about
  - checking the patient in/out
  - Test kit coordination
  - Immediate GC needs or questions

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The GC monthly schedule detailing who is covering MFM clinics in-person or virtually is available. A copy is currently in the MFM Teams group under "Genetic Counseling in MFM" in files and is continuously updated.

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