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| **Time of Appointment:** 2:00 – 2:30 PM | New Patient Est. Patient |  |

**Patient Details**

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| **Name:** Fulton, Olivia | **Age:** 28 y/o | **Biological Sex:** Female | **Gender:** Female |
| **Gravida/Para:** G2P1101 | **Current GA:** N/A | **Delivery:** 256 |  |

**Referral/comments:**

* **Indication Summary:** Patient returns for follow up to discuss fetal autopsy and fetal genetic test results.TOF confirmed on US at 195. Patient was counseled at her prior visit about the available prenatal screening and diagnostic testing options. She declined amniocentesis and cf DNA at her prior appointment. Patient experienced spontaneous loss at 256. Following loss, patient elected for and autopsy and genetic testing on the fetus; results have returned.
* **Relevant Family History:** Patient’s first-born child was delivered vaginally at 381. The child is reportedly healthy with no known congenital anomalies or developmental delays. Family history unremarkable for infertility, others with heart defects, stillbirths, multiple miscarriages, sudden deaths, or known genetic conditions.  No consanguinity reported.

**Prior test results and evaluations:**

* First trimester serum screen with US at 125 weeks – negative
* NT measurement at 125 -78th percentile
* Anatomy scan at 192 identified probable heart defect (Tetralogy of Fallot)
* Targeted US at 195 confirmed TOF, no other anomalies identified
* Fetal ECHO at 241
* Patient evaluated at 251 weeks for decreased fetal movement-no heart tones
* Pregnancy loss at 256
* Elected fetal autopsy and genetic studies (microarray, chromosomes, and exome sequencing)
* Exome sequencing identified a *CHD7* pathogenic variant – CHARGE syndrome
* Microarray and karyotype negative
* Fetal autopsy findings: Tetralogy of Fallot, eye coloboma, TE fistula

**Appointment plan:**

* Educational Counseling:
  + Discuss genetic test results and autopsy findings
  + Natural history of CHARGE Syndrome, incidence, recurrence risk; option for prenatal evaluation
* Psychosocial counseling
  + Assess patient’s experience with her recent loss, provide resources for pregnancy loss, navigating grief, and support communities