

## EVALUATION OF THE ANOMALOUS FETUS/STILLBIRTH

### General

In developed countries, up to 1/5 of all stillbirths can be attributed to fetal disorders. In order to assist the parents in their grief process, an attempt to determine an etiology of their loss should be made. Identifying an etiology will perhaps make the loss more understandable and is critical for counseling the couple about recurrence risks.

In general, the essential components for evaluation of an anomalous fetus include a detailed obstetric and family history, cytogenetic studies, external exam of the fetus, and autopsy. It should be noted that some evaluations or tests might not be paid for by insurance. Careful consideration should be given to requesting the most appropriate tests/evaluations with the highest potential yield.

### Detailed Obstetric, Maternal, and Family History –

- Maternal medical conditions – seizure disorder, diabetes, autoimmune disease, etc
- Teratogen exposures
- Recurrent miscarriages or infertility
- Previous child with anomalies
- Family history of genetic condition, congenital anomaly, developmental delay, etc

### Cytogenetics – a cytogenetic abnormality is identified in up to 20% of dysmorphic fetuses

- Antenatal amniocentesis for karyotype and viral and bacterial culture
  - May provide the best opportunity for cell growth and results
- Postnatal samples should be collected on every anomalous fetus
  - Cord blood, umbilical cord section, or placenta samples are best
    - Use sterile technique (gloves, betadine prep, etc.) when collecting specimens and obtain as quickly as practical
    - Collect 3-5 cc of cord blood in 1) a green top, sodium heparin tube and 2) a purple top EDTA tube
    - Placenta or cord section in cytogenetic tissue transport media
      - 1 x 1cm placenta, from below cord insertion site
      - 1.5cm length cord section
    - Samples can be kept with fetus, up to 48hrs, at room temperature
      - Do not freeze
      - Best to keep sample with fetus until dysmorphology exam is done and determination of additional genetic testing is made
  - The on-call geneticist or pathologist can be contacted with questions

### External Exam/Dysmorphology Exam –

- Brief description of the fetus
  - To be provided by the physician present at delivery
    - Include in the delivery note and mother's discharge summary
    - Photographs if family consent is obtained
- Page the on-call dysmorphologist and document the time of the page

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- In cases when a dysmorphology exam cannot be performed within 48 hours, the fetus should be sent to OMI

**Autopsy** – provides the greatest yield; identifies new information that influences diagnosis and counseling in up to 50% of cases

- Consent for autopsy should be obtained from the parents and the OMI Death Packet filled out
  - Parents can consent to full autopsy or limited
  - Evaluation can involve as little as x-rays/CT scan
    - Radiographic exam is indicated for all suspected skeletal dysplasias
- Fetus should be sent to OMI after dysmorphology exam has been completed or after 48hrs
- The placenta should stay with the stillborn
  - Do not freeze
- *There is no charge to the family for a hospital autopsy at UNM*

**Maternal Studies**

- Appropriate specimens will depend on the clinical history, but in general, certain studies are recommended:
  - Kliehauer-Betke
  - Indirect Coombs
  - Hbg A1C
  - Toxicology screen
  - IgM and IgG titers for CMV, parvovirus B19, toxoplasmosis
  - Anticardiolipin ab, lupus anticoagulant

**Follow-up** – (also see the MFM Protocol on Management of Perinatal Grief)

- Referral to grief counseling services.
- Communication of results
  - Should occur immediately as results are completed
  - Referral to the genetic counselor, primary obstetric care provider, or MFM clinic
    - 6-week post-partum visit
    - Separate referral specifically for results discussion and recurrence risk counseling

**CONSULTATION:** Twenty-Four hour consultation is available by calling the Maternal Fetal Medicine service at the University of New Mexico Hospital, 1-888-866-7257.

**References:**

Management of stillbirth. ACOG Practice Bulletin No. 102. American College of Obstetrics and Gynecologists. Obstet Gynecol 2009; 113:748-61.

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