What do physicians and other caregivers need to know about the Trisomy 13 and 18 genetic variations in the context of pregnancy, infancy, and childhood?

Background

- The prevalence of Trisomy 18 is estimated as 1/6000-1/8000 live births and about 1/8000-1/15,000 live births for Trisomy 13. The overall prevalence is higher (1/2500-1/5000) due to the high frequency of fetal loss and pregnancy termination after prenatal diagnosis.
- Possible perinatal outcomes range widely, from fetal demise to stillbirth to livebirth.
- Many liveborn infants survive to discharge, with and without medical interventions immediately after birth. Even with presumed life-threatening conditions, some infants can be discharged home. (see Appendix)
- Many infants with Trisomy 13 and 18 can successfully undergo surgery. For those infants, selected surgical procedures *may* be helpful in meeting the goals of the family.
- Infants who are discharged home may live for prolonged periods, especially if parents choose medical interventions. Infants of parents who choose ongoing medical interventions will likely require multiple (and sometimes lengthy) re-admissions to the hospital in case of viral infection, to provide further interventions or procedures, or for medical management of worsening symptoms.
- Infants who live into childhood have profoundly delayed development AND:
    - Children are usually described as 'happy' by their parents.
    - Children may recognize family members, smile, and laugh.
    - Children may sleep independently, feed themselves, play independently, understand simple words and phrases, point at objects and follow commands.
    - Some may say "mama" or "papa" and some can learn to use a sign board to communicate.
    - Some children can sit without assistance, some children can crawl and more rarely, some can learn to walk with a walker

Pregnancy

- What expectant parents want when facing a *new* prenatal diagnosis of T13 or 18:
    - Clear, direct communication, in person if possible

- Basic facts about how the diagnosis ~~will~~ may affect the pregnancy and the infant, if born alive
- Choices, options and information about the timing of choices clearly explained in a sensitive manner using neutral language
- Support for the decisions that are made
- Relief from self-blame

Delivery and Infant Care

- What most parents want who have or are preparing to deliver a child with T13 or 18:
  - Acknowledgement that their child is a unique person, not a diagnosis or collection of problems.
  - Individualized counseling: every child has a unique set of challenges and each family has a unique set of values and preferences.
  - Neutral language. (See neutral language guide)
  - A consistent message from all caregivers: therefore communication and consensus among caregivers is crucial. (See Shared Decision-Making)
  - No judgment from health care providers regarding their regard for their child's place in their family.
  - No judgment from health care providers regarding the preferences they express for medical interventions or for comfort care only.
  - An assumption that they have become acquainted with their child's condition over time. For instance, do not begin encounters by saying, "Don't you know your infant is going to die soon?"

    From a parent: "When Lily was 6 months, I took her to the emergency room because she had a virus and was struggling to breathe. The doctor I initially talked to hardly seemed to hear me - he just kept staring at her and asking me if I was sure she had Trisomy 18 because if so, she wasn't supposed to be alive. Lilly had two hospital stays for viruses and both times, several doctors asked if we knew that she wasn't going to live much longer. Since there are adults - in their 30s! - with Trisomy 18 this is a very upsetting to say to their parents!"
  - Respect for the hopes they have for their child. For instance, from a parent: "I understand doctors wanting to share facts …. but please, remember that parents also need HOPE."
    - Prior to delivery, expectant parents often hope for many things such as:
      - A live birth so they can meet their child alive.
      - To introduce their child to his or her family, to create memories, and celebrate a new family member
      - To take their child home.
    - When infants are discharged home after birth, families generally hope
      - To give their child a good and meaningful life, loving their child and being a family for as long as possible.