

A young boy with short brown hair is standing on a grassy hill. He is wearing a bright orange zip-up jacket and dark blue jeans. He is looking directly at the camera with a slight smile. The background is a clear, solid blue sky. The grass at the bottom of the frame is green and slightly out of focus.

DELIVERING A PRENATAL
OR POSTNATAL DIAGNOSIS

Communicating a prenatal or postnatal diagnosis to a family can be overwhelming for both the clinician and the patient. That moment is often described as a flashbulb memory that a patient remembers in detail for a lifetime. Fortunately, best practice recommendations outline suggestions for discussing a prenatal or postnatal diagnosis so that clinicians can frame that moment in sensitivity and compassion (Skotko et al., 2009; Sheets et al., 2011). In addition, resources are available to help clinicians along the way.

PRENATAL DIAGNOSIS RECOMMENDATIONS

Best practice guidelines for communicating a prenatal diagnosis include the following recommendations (Skotko et al., 2009):

- Clearly outline the differences between prenatal screening and diagnostic tests. Importantly, patients need to understand that screening tests (including cell-free DNA and non-invasive prenatal screening tests) indicate a patient's chances for having a baby with a number of genetic conditions. However, the screening tests are not definitive because false positives do sometimes occur. Only chorionic villus sampling and amniocentesis are considered diagnostic. (Gregg, 2013)
- Ask the pregnant woman about why having a diagnosis prior to birth would be important to her to better guide any future conversations about a diagnosis.
- When possible, deliver the results in person or at a pre-established time by phone. Determine a standard way of handling all results and tell patients about that up front so that they don't get the impression that an appointment or phone call is only scheduled if results indicate a diagnosis.
- Personally deliver the diagnosis as soon as possible following definitive prenatal testing. Use commonly understandable terms and convey information in a patient's native language, when translation is available.
- Each condition detected with prenatal testing has different outcomes, and each expectant parent reacts differently based on his or her background and experience, life circumstances, and perceptions about parenting. Assess the emotional reactions of the expectant parents, and validate these feelings. Use active listening and empathetic responses to offer support. (Sheets et al., 2011)
- If a condition does not cause premature death, use neutral language such as, "The results indicate..." and not begin with, "I'm sorry," or "Unfortunately, I have some bad news..."
- Provide accurate and up-to-date information about the genetic condition and contact information for local support organizations.

POSTNATAL DIAGNOSIS RECOMMENDATIONS

Best practice guidelines for communicating a postnatal diagnosis include the following recommendations (Skotko et. al., 2009):

- Deliver the news in person to new parents using commonly understandable terms and convey information in a patient's native language, when translation is available.
- Discuss the diagnosis in a private setting with the parents as soon as you can after confirming the results.
- Inform both parents together, if possible.
- Each condition has different outcomes, and each parent reacts differently based on his or her background and experience, life circumstances, and perceptions about parenting. Assess the emotional reactions of the expectant parents, and validate these feelings. Use active listening and empathetic responses to offer support. (Sheets et al., 2011)
- "Allow time for silence and time for tears. Do not feel that you need to talk to 'fill the silence.' Offer the family time alone." (Sheets et al., 2011)
- If a condition does not cause premature death, avoid beginning a conversation with language that conveys pity or sorrow, and start with positive language, such as, "Congratulations" or "Your baby is beautiful, and we suspect..."

- Provide accurate and up-to-date information about the genetic condition and contact information for local support organizations.

PATIENT EDUCATION

Experts recommend that up-to-date information on genetic conditions should be available to new and expectant parents whenever screening or diagnostic results suggest or confirm a diagnosis. They emphasize that parents should be informed about the following:

- Genetics of the condition
- Available support services, such as Early Intervention
- Range of health conditions seen in infants and young children with the condition and life expectancy
- Range of life outcomes including common strengths and challenges based on current data
- List of resources that also includes family and psychosocial outcomes
- Photographs (Levis et al., 2012)

All of these criteria are met by the National Center for Prenatal and Postnatal Resources (lettercase.org) materials which are reviewed by representatives of the national medical and disability organizations. (Levis et al., 2012) The resources are available in multiple languages about different genetic conditions.

RESOURCES

Because research, education, opportunities, and healthcare for individuals with different genetic conditions are constantly evolving, it is important to provide patients with references to genetic counselors who are trained to provide more information, as well as reliable local and national patient support groups (ACOG, 2007). The national patient advocacy organizations and many local patient advocacy groups also offer prenatal and new parent support programs, as well as medical outreach programs and resources.

Adoption services, such as the National Down Syndrome Adoption Network (ndsan.org), are also available for certain condition groups and maintain registries of families interested in adopting infants with different conditions.

National Center for Prenatal and Postnatal Resources/Lettercase › lettercase.org

Brighter Tomorrows › brightertomorrows.org and brighter-tomorrows.org (for medical professionals)

MassGeneral Hospital Academy webinar on prenatal testing by Dr. Brian Skotko (with CME credit) › mghacademy.org/downsyndrome

Health recommendations for children with different genetic conditions › healthychildren.org

Genetic Support Foundation › geneticsupportfoundation.org

Perinatal Quality Foundation › perinatalquality.org

National Society of Genetic Counselors › nsgc.org

American College of Genetics and Genomics › acmg.net

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