Conversations About Down Syndrome

Delivering and discussing a Down syndrome diagnosis

Presented by

 Disclosure

• I have nothing to disclose.

• My husband, Chris Small, works at Genentech/Roche. He is not involved in marketing their drugs; he edits the submission materials that go to the FDA.

Our Goals

• To help healthcare professionals understand supportive, informative ways to discuss Down syndrome with families

• To inform healthcare professionals about local and national resources available to support and enhance the lives of people with Down syndrome

• To highlight specific medical and developmental needs of people with Down syndrome

Down Syndrome 101

• Most common genetic condition, occurring in 1:691 births in the United States

• Estimated 6,000 births in the U.S. per year

• More than 200,000 Americans have Down syndrome

• Present in all races and socioeconomic classes and is equally common in both genders

• No known cause for Down syndrome—it is due to extra genetic material in the egg or sperm and is present from the moment of conception
Types of Down Syndrome

- **Trisomy 21**
  - Most common type, accounting for 95% of all cases of Down syndrome
  - An additional copy of chromosome 21 is replicated in every cell of the body
- **Translocation**
  - Accounts for 4% of Down syndrome cases
  - Part of chromosome 21 breaks off during cell division and attaches to another chromosome (often chromosome 14)
- **Mosaicism**
  - Accounts for 1% of Down syndrome cases
  - Additional copy of chromosome 21 is replicated in only some cells, so the signs of DS may present differently than in cases of Trisomy 21 or translocation.

Physical Characteristics

Each person with Down syndrome will have unique physical characteristics, but common ones seen in people with Down syndrome include:

- Hypotonia (low muscle tone)
- Epicanthal folds on inner corners of eyes (with an almond shape)
- Flat nasal bridge
- Small or unusually shaped ears
- Excess skin at the back of the neck
- Tongue that protrudes or seems large compared to size of mouth
- White spots on colored part of the eye (called “Brushfield spots”)
- Single, deep crease across the palm of the hand
- Extra space between first and second toes (called “sandal gap”)

Medical Concerns in Babies and Young Children

Some babies with Down syndrome are born with no other medical concerns. However, certain medical conditions can be somewhat common in children with Down syndrome:

- Congenital heart problems (50%)
- Hearing problems
- Obstructive sleep apnea
- Gastrointestinal atresia or Hirschprung’s disease
- Thyroid dysfunction (especially hypothyroidism)
- Leukemia
- Nursing and feeding difficulties

Medical Concerns in Older Children and Adults

- Atlantoaxial instability
- Vision problems
- Autism
- Precocious puberty
- Early-onset menopause
- Celiac disease
- Dementia and Alzheimer’s disease

The AAP provides guidelines for recommended screenings at a child’s well-baby/well-child checks. Down Syndrome Connection can provide these guidelines, too.
**Diagnosis**

- Diagnosis can happen prenatally, at birth, or occasionally several months later.
  - Birth diagnoses may be suspected but cannot be confirmed until the results of a karyotype test come back.
  - If a family asks what type of Down syndrome their child has before the karyotype has confirmed, it is best to prepare them for Trisomy 21, as that is by far the most common type.
- Every parent will react to the news differently, but most follow the general cycle of:
  - Grief > Denial > Bargaining > Anger > Acceptance
- Regardless of how/when a baby is diagnosed, the birth should be a positive event, and the baby’s arrival should be celebrated.

**Prenatal Diagnosis**

Although many parents don’t receive a suspected diagnosis until their baby is born, women are increasingly choosing to do noninvasive prenatal screening tests such as Harmony and MaterniT21, which can indicate the possibility of a chromosomal abnormality, such as Down syndrome.

- These tests are _screens_, not definitive tests. Results are not 100% accurate. There may still be delivery-room surprises.
- A majority of women receiving a “positive” indication on these screens choose to end their pregnancies. However, even if they continue the pregnancy and come to terms with the diagnosis while pregnant, the delivery room is still a very emotional, raw place for them.

**What Can You Do?**

- Treat the baby’s arrival as the celebration that it is. A _baby_ was brought into the world!
- Congratulate, don’t apologize. Most parents report that it hurt to have people not congratulate them on the birth of their baby.
- Use the baby’s name.

**What Can You Do?**

- Use “people first” language. It is a baby with Down syndrome, not a “Down’s baby” or “Down syndrome baby.”
- Tell both parents together.
- Remember that mothers and fathers can respond differently to the news.
What Can You Do?

- Answer questions honestly and knowledgeably. If you don’t know an answer, tell the parents you will find out—and then contact us, and we’ll help you.
- Allow parents time to bond with their child, but check in regularly to see whether parents need anything.
- Recognize that certain statements may seem judgmental to parents, such as, “Did you have prenatal testing?”
- Be aware of the medical issues that can come with Down syndrome and answer parents’ questions honestly, but also emphasize that many children with Down syndrome experience none of these conditions and that most conditions are very treatable.
- Trust parent intuition. Check further into any concerns before assuming that something is “just part of Down syndrome.”

What Can You Do?

- Provide referrals to Down Syndrome Connection of the Bay Area and to the Regional Center for early intervention.
  - Social Services should have contact information for both.
  - DSCBA will deliver a welcome basket with gifts and further information for parents.
  - DSCBA can provide a parent-to-parent match.

What Is Early Intervention?

- Children with Down syndrome are entitled to receive early intervention services through the Regional Center.
- Services include infant development, occupational therapy, physical therapy, and speech therapy.
- Down Syndrome Connection works in conjunction with the Regional Center, providing support and enrichment where early intervention leaves off.
- California Department of Developmental Services provides a full list of area Regional Centers on their website.

What Is Early Intervention?

- Children with Down syndrome are entitled to receive early intervention services through the Regional Center.
- Services include infant development, occupational therapy, physical therapy, and speech therapy.
- Down Syndrome Connection works in conjunction with the Regional Center, providing support and enrichment where early intervention leaves off.
- California Department of Developmental Services provides a full list of area Regional Centers on their website.

Quality of Life

Worries about a person with Down syndrome’s quality of life are unfounded, as are concerns that the person will be a “burden” to his or her family. According to large studies of people with Down syndrome and their families:

- 99% of people with DS indicated they are happy with their lives, and 97% like who they are.
- 86% indicated that they make friends easily, and those who struggled tended to have isolated living situations.
Expectations

- Average lifespan for a person with Down syndrome is about 60–65 years.
- Every person with Down syndrome has unique cognitive, social, and physical abilities, as well as unique behavior and accomplishments.
- More and more children with Down syndrome are becoming fully included in school, enhancing their success.
- Adults with Down syndrome can:
  - Have relationships and marry
  - Attend post-secondary schools
  - Live semi-independently or independently
  - Work at rewarding jobs
  - Actively participate in their community
- Each person with Down syndrome is unique, and there’s no way to know what the future holds.

Common Myths

- Only women older than age 35 are likely to have a baby with Down syndrome.
  - False! 80% of babies with Down syndrome are born to women under the age of 35.
- A child with Down syndrome will have a negative effect on his or her parents.
  - False! 99% of parents report loving their child with DS. And the divorce rate in families who have a child with DS is actually lower than in families with a nondisabled child.
- A child with Down syndrome will have a negative effect on his or her siblings.
  - False! 97% of siblings of people with DS reported being proud of their sibling with DS. And 88% of siblings of people with DS reported that they think they are better people for having a sibling with DS.

Common Myths

- People with Down syndrome are always happy.
  - False! People with Down syndrome experience the same range of emotions as anyone else. Anecdotally, though, people with Down syndrome do tend to often have a largely positive outlook on life.
- “God only gives special children to special people.”
  - Many parents—even those with a strong faith—feel offended by this statement. In their minds, they are normal, everyday parents who just happened to have an exceptional child.

What DSCBA Does

- One-on-one parent mentors for new families
- Support groups for parents, grandparents, and other family members
- Developmental-based classes for infants through adults
- School-readiness summer program
- Communication technology services
- On-site reading teacher
- Music therapy for infants and children
- On-site feeding/oral-placement therapist
- Special events throughout the year, such as seminars, training sessions, camps, etc.
- Medical Outreach Alliance (partnering with 37 area hospitals)
- Education Alliance (partnering with 32 Bay Area school districts)
What DSCBA Doesn’t Do

• Provide early intervention services. That is provided by the Regional Center.
• Turn anyone away. Services are inclusive for everyone, and we welcome family members of people with Down syndrome to be a part of our community.

Other Resources

• Global Down Syndrome Foundation
• LuMind Foundation
• National Down Syndrome Congress
• National Down Syndrome Society
• Down Syndrome Diagnosis Network
• Local family resource networks
• Center for Down Syndrome at Lucile Packard Children’s Hospital
• Charlie’s Clinic at UCSF Benioff Children’s Hospital Oakland
• Regional Center for early intervention

References

• 1 Centers for Disease Control and Prevention (http://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html)