

Down Syndrome in a Newborn

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Key Points

- While maternal serum screening and prenatal ultrasound identify the majority of pregnancies at increased risk for Down syndrome, some cases are missed.
- Newborns with Down syndrome must be evaluated at birth for specific medical complications.
- Forthright presentation of the news that a child has Down syndrome, including a balanced presentation of the condition, respect for parents' privacy, opportunities for parents to ask questions, and ready availability of follow-up, is associated with greater parental satisfaction.

Learning Objectives

Participants will be able to:

- List the major medical complications in newborns with Down syndrome;
- Explain how the karyotype of an affected child influences risk assessment for the parents' future pregnancies.

Family History Issues

- Most commonly, Down syndrome occurs as trisomy 21, i.e., the presence of three separate copies of chromosome 21. This is a sporadic condition and other family members are unlikely to be affected.
- In about 3% of cases, Down syndrome occurs as a result of an unbalanced translocation involving chromosome 21, a condition which can be inherited. See [Risk Assessment](#) below.

Red Flags



Some of the physical features leading to the suspicion of Down Syndrome at birth include:

- Hypotonia
 - A small brachycephalic head
 - Epicanthic folds
 - Flat nasal bridge
 - Upward slanting palpebral fissures
 - Brushfield spots of the iris (grey-white specks of depigmentation seen in a ring around the pupil)
 - Small mouth
 - Small ears
 - Excessive skin at the nape of the neck
 - Single transverse palmar crease
 - Wide space between the first and second toes
 - Short fifth finger with clinodactyly
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Case 22. Down Syndrome in a Newborn

Mrs. P, a 31-year-old woman, had an uneventful first pregnancy. At 16 weeks' gestation, a triple screen blood test was normal, indicating a 1/275 risk of Down Syndrome; based on this test result, prenatal diagnosis was not considered. An ultrasound examination performed at 18 weeks' gestation revealed no abnormalities. Mrs. P's family history is significant for a 12-year-old maternal first cousin with Down syndrome.

At birth, Mr. and Mrs. P's infant daughter was noted to have hypotonia, a heart murmur, and facial features suggestive of Down syndrome. Mrs. P's physician met with her and Mr. P to discuss the suspicion that the infant had Down syndrome. Blood was obtained then for chromosomal studies; one week later, trisomy 21 (47, XX, +21) was confirmed.

Clinical Care Issues

Maternal serum screening for Down syndrome

While maternal serum screening can detect approximately 60%-75% of cases of Down syndrome, some cases are missed. The triple screen has a Down syndrome detection rate of approximately 60% with a false positive rate of 5%; the quad screen has a detection rate of about 70% with a false positive rate of 5%. First trimester screening (maternal serum tests +

ultrasound measurement of nuchal translucency) can detect about 75% with a false positive rate of 5%.

The triple screen risk of 1/275 calculated for Mrs. P was somewhat higher than her age-related risk of approximately 1/826. Still, her results did not "screen positive" as determined by the reference laboratory (laboratories typically use a cut-off for "screen positive" that is equal to the risk of a 35-year-old woman having a baby with a chromosome abnormality, or approximately 1/200.)

Clinical evaluation of the newborn

Newborns with Down syndrome should be evaluated for specific complications including:

- Congenital heart defects (50% risk)
- Bowel obstruction (typically duodenal atresia, about 12% risk) usually evident by persistent vomiting
- Hearing loss (60% risk)
- Ocular abnormalities (congenital cataracts, strabismus)
- Constipation with an increased risk of Hirschsprung disease (<1% risk)
- Leukemia (1% risk); polycythemia (18% risk)
- Congenital hypothyroidism (1% risk)

Risk Assessment

Several screening methods are available during pregnancy to detect an increased risk of Down syndrome (see [Case 21, Risk Assessment](#)).

After the birth of a child with Down syndrome, the risk of Down syndrome in a future pregnancy is determined by reviewing the chromosome report(s) and by considering maternal age at the time of a future pregnancy. If the chromosome report indicates trisomy 21, as is the case for Mr. and Mrs. P's child, the future risk is relatively low, but when Down syndrome is due to a chromosomal translocation, the risk may be significant. Reviewing the blood chromosome report in this case was particularly critical because Mrs. P has a relative with Down syndrome. This positive family history raised the index of suspicion for an inherited unbalanced translocation.

Down syndrome due to translocation. In about 3% of cases of Down syndrome, the condition occurs as a result of an unbalanced translocation between chromosome 21 and another chromosome. In such cases, the

individual with Down syndrome has an additional chromosome 21 that is attached to another chromosome (typically chromosome 13, 14, or 15). One of the parents could be carrying a balanced translocation, which means that one of the two chromosome 21's is attached to the other chromosome, but there is no extra or missing genetic material. In that case, the parent has an increased risk of having another child with Down syndrome or passing the balanced translocation to their offspring who would also be at risk of having a child with Down syndrome. (See diagrams of Robertsonian translocations: [Genetics & Public Policy site](#), [Centre for Genetics Education](#).) Therefore, if a baby has translocation Down syndrome, a chromosome study should be performed on both parents. Genetic counseling and discussion of testing of other at-risk family members should be offered.

Risk in this case (trisomy 21). Mr. and Mrs. P's baby has trisomy 21, which is sporadic and non-inherited. Therefore, Mr. and Mrs. P do not need to have chromosome studies. If Mr. and Mrs. P have more children in the near future, their risk of having another child with a chromosome abnormality is 1% since that is the empiric risk for all couples who have had one child with a trisomy. However, once Mrs. P is age 38 years, the risk of having a child with a chromosome abnormality becomes her maternal age-related risk and increases as her age increases. Other family members are not at increased risk of having a child with a chromosome abnormality, and the fact that her cousin has Down syndrome does not further increase her risk.

Genetic Counseling and Testing

The family can be offered a genetics consultation to discuss Down syndrome in more detail. A genetics consultation includes a review of the overall health of the child, discussion of the medical implications of Down syndrome, and guidance to the family as they plan for follow-up evaluations with medical specialists. In addition, the genetic counselor's role is to further discuss the genetics of Down syndrome, the implications of the diagnosis, and to support parents in their coping efforts. Some parents may welcome the opportunity to meet and talk to other parents of children with Down syndrome or to join a support group. Genetic counselors can facilitate access to resources including books, articles, or Web sites.

Interventions

Comprehensive guidelines for the care of children with Down syndrome are available [[Cooley 1991](#), [AAP 2001](#)]. Recommendations include evaluating the

newborn for complications listed above as well as monitoring children for hearing and vision problems, respiratory and ear infections, hypothyroidism, atlantoaxial instability, and other problems observed in children with Down syndrome. See guidelines for extensive information.

All individuals with Down syndrome have some degree of cognitive impairment. Most have mild to moderate mental retardation, but severe mental retardation can occur. Enrollment in an early intervention program should occur shortly after birth. Early intervention includes physical therapy, speech, and occupational therapy and helps in each of the four main areas of development: gross motor and fine motor skills, language, social development, and self-help skills. Programs are individualized to help each child in every area of development.

Ethical/Legal/Social/Cultural Issues

Breaking the news

Studies have found that parents are generally dissatisfied with the delivery of the news or the suspicion that their newborn has Down syndrome [Cunningham et al 1984]. Although it could be argued that receiving this information is always a difficult experience, a "model service" has been developed from parents' feedback that was tested and found to have 100% satisfaction [Cunningham et al 1984]. The following practices were incorporated into the model service:

- Tell the parents as soon as possible.
- Have a person with sufficient knowledge provide the initial information.
- Have both parents and the baby together.
- Tell the parents in a private place.
- Be straightforward.
- Use clear language.
- Provide time to ask questions.
- Provide a balanced point of view of the condition.
- Plan for follow-up discussion.
- Provide phone numbers for a contact person for questions.
- Make other information sources available.
- Let the parents have uninterrupted private time with their child after the discussion.

Resources

- **National Down Syndrome Society Web Site**
- **National Down Syndrome Congress**
- **National Library of Medicine Genetics Home Reference**
[Down syndrome](#)
- **March of Dimes: Down Syndrome**
- **Family Voices**
- **Genetic Alliance**
- **GeneTests Online Medical Genetics Information Resource**

References

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