

Health Reference Series

Fifth Edition

Genetic Disorders

SOURCEBOOK



Basic Consumer Health Information about Heritable Disorders, Including Disorders Resulting from Abnormalities in Specific Genes, Such as Hemophilia, Sickle Cell Disease, and Cystic Fibrosis, Chromosomal Disorders, Such as Down Syndrome, Fragile X Syndrome, and Klinefelter Syndrome, and Complex Disorders with Environmental and Genetic Components, Such as Alzheimer Disease, Cancer, Heart Disease, and Obesity

Along with Information about the Human Genome Project, Genetic Testing and Newborn Screening, Gene Therapy and Other Current Research Initiatives, the Special Needs of Children with Genetic Disorders, a Glossary of Terms, and a Directory of Resources for Further Help and Information

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Chapter 66

When Your Baby Has a Birth Defect

We see happy images of and tend to hear about only healthy babies. But many babies are born with a birth defect. These are abnormalities of structure, function, or body chemistry that will require medical or surgical care or could have some effect on a child's development.

About 150,000 babies are born in the United States each year with birth defects, according to the March of Dimes. There is a wide range of birth defects, from mild to severe, and they can be inherited or caused by something in the environment. In many cases, the cause is unknown. Doctors may detect a birth defect during prenatal testing.

If you've just found out that your child has a birth defect, you're probably experiencing many emotions. Parents in your situation often say they feel overwhelmed and uncertain about whether they'll be able to care for their child properly.

Steps to Take

Fortunately, you aren't alone—you'll find that many people and resources are available to help you. As the parent of a child with a birth defect, it's important for you to:

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- **Acknowledge your emotions:** Parents of children with birth defects experience shock, denial, grief, and even anger. Acknowledge your feelings and give yourself permission to mourn the loss of the healthy child you thought you'd have. Talk about your feelings with your spouse or partner and with other family members. You might also consider seeing a counselor. Your doctor might be able to recommend a social worker or psychologist in the area.
- **One of the best things you can do for yourself and your child is to seek support:** Getting in touch with someone who's been through the same thing can be helpful; ask your doctor or a social worker at your hospital if there are other parents in the area who have children with the same condition. Joining a support group may also help—ask the doctors or specialists for advice on finding a local or national support group or search online.
- **Celebrate your child:** Remember to let yourself enjoy your child the same way any parent would—by cuddling or playing, watching for developmental milestones (even if they're different from those in children without a birth defect), and sharing your joy with family members and friends. Many parents of kids with birth defects wonder if they should send out birth announcements. This is a personal decision—the fact that your child has a health problem doesn't mean you shouldn't be excited about the new addition to your family.

Getting Help and Information

Seek information: The amount each person would like to learn varies from parent to parent, but try to educate yourself as much and as soon as you are able. Start by asking your doctors lots of questions. Record the answers as best you can. If you're not satisfied with the answers—or if a doctor is unable to answer your questions thoroughly—don't be afraid to seek second opinions.

Other places to get information include:

- books written for parents of children with birth defects;
- national organizations such as the March of Dimes, the National Information Center for Children and Youth With Disabilities, or those representing a specific birth defect;
- support groups or other parents.

When Your Baby Has a Birth Defect

Keep a file with a running list of questions and the answers you find, as well as suggestions for further reading and any materials your child's doctor gives you. In addition, keep an updated list of all health care providers and their phone numbers, as well as emergency numbers, so you're able to reach them quickly and efficiently.

Part of this process of collecting information should involve exploring options for paying for treatment and ongoing care for your child. There can be extra medical and therapeutic costs associated with caring for a child with a birth defect. Besides health insurance, other resources are available, including nonprofit disability organizations, private foundations, Medicaid, and state and local programs. One of the hospital social workers should be able to help you learn more about these.

Seek early intervention: Early intervention is usually the best strategy. Designed to bring a team of experts together to assess your child's needs and establish a program of treatment, early intervention services include feeding support, identification of assistive technology that may help your child, occupational therapy, physical therapy, speech therapy, nutrition services, and social work services.

Besides identifying, evaluating, and treating your child's needs, early intervention programs will:

- tell you where you can get information about the disability;
- help you to learn how to care for your child at home;
- help you determine your payment options and tell you where you can find services for free;
- help you make important decisions about your child's care;
- provide counseling.

Your child's doctor or a social worker at the hospital where you gave birth should be able to connect you with the early intervention program in your area.

Use a team approach: Most children with birth defects require a team of professionals to treat them. Even if your child needs to see only one specialist, that person will need to coordinate care with your primary doctor. Although some hospitals already have teams ready to deal with problems such as heart defects, cleft lip and palate, or cerebral palsy, you may find yourself having to serve as both the main point of contact between the different care providers and the coordinator of your child's appointments. As soon as you are able, get

to know the different team members. Make sure they know who else will be caring for your child and that you intend to play a key role.

The Future of Birth Defects

Research into the environmental and genetic causes of birth defects is ongoing. Technology contributes to understanding and preventing defects in various ways—for example, prenatal testing is growing increasingly sophisticated.

Safer and more accurate tests include:

- results of ultrasound tests and magnetic resonance imaging (MRI), which are sometimes combined with information from blood tests to determine the risk of having a child with certain birth defects;
- maternal blood screening to determine risk of chromosomal abnormalities;
- amniocentesis and chorionic villi sampling;
- pre-conception counseling to help you understand any risks for having a child with a birth defect.

Although none of these tests can prevent birth defects, they give a clearer, safer, and more accurate diagnosis at an earlier stage of pregnancy—giving parents more time to seek advice and consider their options.

Genetics research is advancing quickly. The Human Genome Project is working on identifying all of the genes in the human body, including gene mutations that are associated with a high risk for birth defects.

Early surgery is becoming an option in the treatment of some birth defects—and can take place even before a child is born. Surgeons now operate on fetuses to repair structural defects, such as hernias of the diaphragm, spina bifida, and lung malformations. These treatments can be controversial, however, because they can cause premature labor. And it's still a bit unclear as to whether they ultimately improve the child's outcome.

To get information on specific research about your child's disability, contact the national organization for that disability. Also, the March of Dimes and the National Information Center for Children and Youth With Disabilities and the National Organization for Rare Disorders, Inc. (NORD) may have information about current research.

Chapter 67

Tips for Parenting a Child with a Disability

If you have a child with a disability, you are not alone. Millions of parents in the United States are raising children with disabilities. Many resources (including fellow parents) can help you along the way. Here are some tips for parents:

- Learn as much as you can about your child’s disability.
- Find programs to help your child.
- Talk to your family about how you’re feeling.
- Talk to other parents of children with disabilities.
- Join a support group.
- Stick to a daily routine.
- Take it one day at a time.
- Take good care of yourself.

An important quality that you will need to nurture in your child is called “self-determination.” Children who develop this quality have a sense of control over their lives and can set goals and work to attain them. Self-determination is important for all children, but researchers have found that students with disabilities who also have high levels of self-determination are more likely to become adults who are:

“Parenting a Child with a Disability,” U.S. Department of Health and Human Services, Office on Women’s Health, September 22, 2009.

- employed;
- satisfied with their lives; and
- living independently, or with support, outside of their family homes.

Here are some tips to help your child become self-determined:

- As early as possible, give your child opportunities to make choices and encourage your child to express wants and wishes. For instance, these could be choices about what to wear, what to eat, and how much help with doing things your child wants from you.
- Strike a balance between being protective and supporting risk taking. Learn to let go a little and push your child out into the world, even though it may be a little scary.
- Guide children toward solving their own problems and making their own choices. For instance, if your child has a problem at school, offer a listening ear and together brainstorm possible solutions. To the extent that your child can, let your child decide on the plan and the back-up plan.

Programs and Services

Every state has programs and services that can help you meet your child's and your family's needs:

- Early intervention services try to address the needs of children with disabilities and the needs of their families as early as possible. Often, the sooner issues are addressed, the better the outcome. Examples include nutrition counseling for parents, physical therapy for a baby with cystic fibrosis, or sign language lessons for a deaf child. Services vary by state.
- Special education and related services ensure that each child is given a free public education that accommodates his or her special needs. The law requires that every student with a disability have an Individualized Education Program (IEP), which is a plan for that child's education. The IEP includes a list of the services, accommodations, and assistive technology your child will need to succeed in school. Parents of a child with a disability are an important part of the team that writes the IEP. To the extent that they can, children with disabilities should also be encouraged to take part in writing the IEP.

Tips for Parenting a Child with a Disability

- Parent Training and Information (PTI) centers provide parents with information about disabilities and legal rights under laws involving children with disabilities. PTIs can also tell you about resources in the community, state, and nation. PTI centers conduct workshops, conferences, and seminars for parents. And many have libraries where you can borrow books and videos. Every state has at least one PTI. Some states also have Community Parent Resource Centers (CPRCs). CPRCs do the same work as the PTIs, but they focus on reaching underserved parents of children with disabilities. Underserved parents include low-income parents, parents with limited ability to speak and write English, and parents with disabilities.
- Parent to Parent is a program that provides information and one-to-one emotional support to parents of children with disabilities. Trained and experienced parents are carefully matched in one-to-one relationships with parents who are new to the program. The matches are based upon similarities in disability and family issues.