



Prenatal DNA Resource

Checklist for Parents

In speaking to parents, we hear how difficult it is to plan the next steps after receiving the diagnosis of a deletion or duplication from prenatal genetic testing (usually from a procedure like amniocentesis or chorionic villus sampling (CVS)). If you are one of these parents, the checklist below can provide guidance for your child's early years:

- **Find the right pediatrician.** Things to think about:
 - Are they willing to work with you and learn about the syndrome?
 - Do they have experience with children who have special needs and neurodevelopmental disorders?
 - Do they listen and respond to your questions and concerns about your child?

- **Consider finding a developmental pediatrician.** These doctors will assess your child's developmental progress, including speech and behavior concerns. Things to think about:
 - Are they willing to work with you and learn about the syndrome?
 - Some of these specialists have waitlists that go out as far as a year, so call as early as you can.

- **Find a genetics specialist in your area.** These doctors have special training in human genetics and can help you understand the complexities of your child's diagnosis. Resources:
 - www.ACMG.net and click on "find genetic services."
 - www.NSGC.org and click on "find a genetic counselor."

- **Keep up with research by registering with prenatalDNAresource.com.** This scientific group can help you:
 - Connect with genetics, pediatrics, and clinical research experts.
 - Involve you in and update you about the newest research.
 - Help you connect with other parents going through a similar experience.



□ **Check into early intervention services offered by your home state.**

Some possibilities include:

- Ask your pediatrician about getting an early intervention appointment.
- <http://ectacenter.org/contact/ptccoord.asp> overviews early intervention services state by state.
- Find reliable information about special education law and advocacy for children with special needs at www.wrightslaw.com

□ **Consider discussing a referral to a medical specialist with your child's regular physician.** This can be helpful if your child has needs

relating to a specific organ system. Some examples include:

- Cardiologist (heart)
- Endocrinologist (the body's hormonal system)
- Orthopedist (skeletal system)

□ **Pubmed.org** can help you find medical research.

- If you find a researcher who is publishing a lot on the topic you are interested in, consider emailing that researcher.

□ **Internet resources:**

- Search Facebook for family groups interested in specific deletions or duplications.
- www.pedstest.com
- www.howkidsdevelop.com
- "Unique" is an international support group and you can find them at www.rarechromo.org
- SWAN (Syndromes Without A Name) can be found at www.undiagnosed-usa.org

□ **Be sure to take care of yourself!**

- Consider setting up a relationship with a therapist or counselor. Check if your health insurance covers this – they often do!
- Schedule and keep your regular check-ups.
- Get appropriate rest and remember that you can best help your family when you are feeling well yourself.