A Balanced Approach to Delivering a Diagnosis of Down Syndrome

What You Say Really Matters

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About the Presenter

Emily Mondschein is the proud mother of two boys, one who was born with Down syndrome. She holds a master's degree in education and has founded two not-for-profits that serve individuals with Down syndrome.

Emily currently serves as the Executive Director for GiGi's Playhouse Down Syndrome Achievement Centers in Buffalo, NY. She is also a leader of and member on multiple councils, both local and national, that support individuals with disabilities.

Emily has advocated to Congress on behalf of individuals with Down syndrome and is a member of NDSS' National Inclusive Education Task Force and DS-Ambassador program. Most recently she worked with Oishei Children's Hospital to launch a Down syndrome program within the Robert Warner, MD Rehabilitation Center and chairs the Parent Advisory Counsel for the program.

She is also working with NDSS to advance inclusive education guidelines for individuals with Down syndrome the federal level and with the NYS legislature to advance Down syndrome-focused bills at the state level.

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Conflict of Interest

I have no actual or potential conflict of interest in relation to this program/presentation.

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Presentation Objectives

By the end of this presentation, learners will be able to:

- Understand best practices around delivering a pre- or postnatal diagnosis of Trisomy 21
- Fully support new and expectant parents as they navigate and learn about Down syndrome
- Find nationally-recommended resources about prenatal testing and Down syndrome

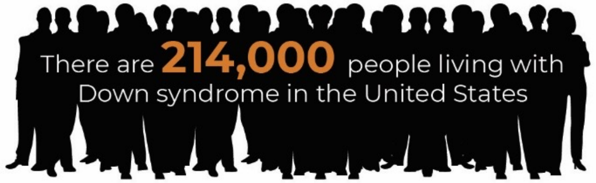
Down Syndrome 101

1. Down syndrome occurs when there are three copies of the 21st chromosome.
2. Down syndrome is the largest chromosomal disability in our country, yet it's the least funded.
3. The average age to have child with Down syndrome is 28.
4. The cause of Down syndrome is unknown, and it does not discriminate across racial or socioeconomic lines.
5. Individuals with Down syndrome have low muscle tone which makes doing everyday skills like walking, talking, eating, reading, etc. much harder than the typical population.
6. 70-80% of people terminate their pregnancy when Down syndrome is suspected. 95-100% termination rate in Iceland and other countries who are intolerant of the diagnosis.

Facts About Down Syndrome

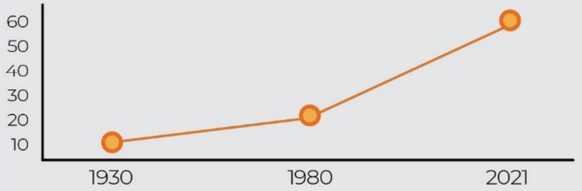
Down syndrome is the most common genetic condition

1 in 768 or **6000**
pregnancies per year



There are **214,000** people living with Down syndrome in the United States

The average **LIFE EXPECTANCY** has increased from 25 years in 1980 to 60 years today



As medical advances continue, life expectancy will continue to increase

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People with Down syndrome possess many strengths and individual talents.

The cognitive impacts of Down syndrome tend to range from **MILD TO MODERATE**

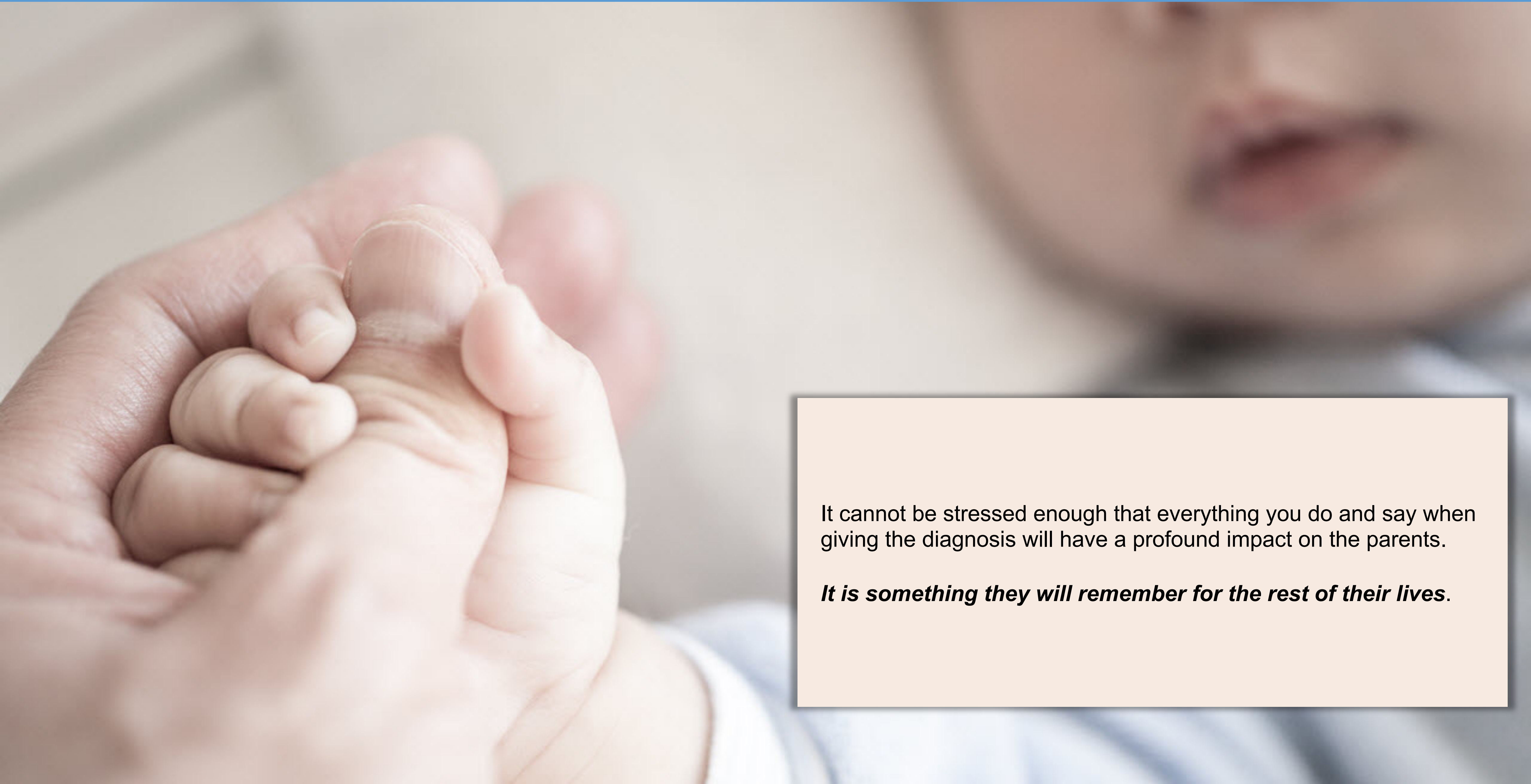
People with Down syndrome are able to meet the same milestones as their peers, just at a slower pace.

Children are able to be part of general education classrooms, continue with higher education, and have jobs within the community.

People with Down syndrome are able to live independently or in group homes, take part in meaningful relationships, and get married.

Government funded programs, such as Early Intervention, are available to help. Children who begin therapies at a young age show significant gains in life skills compared to those who wait.

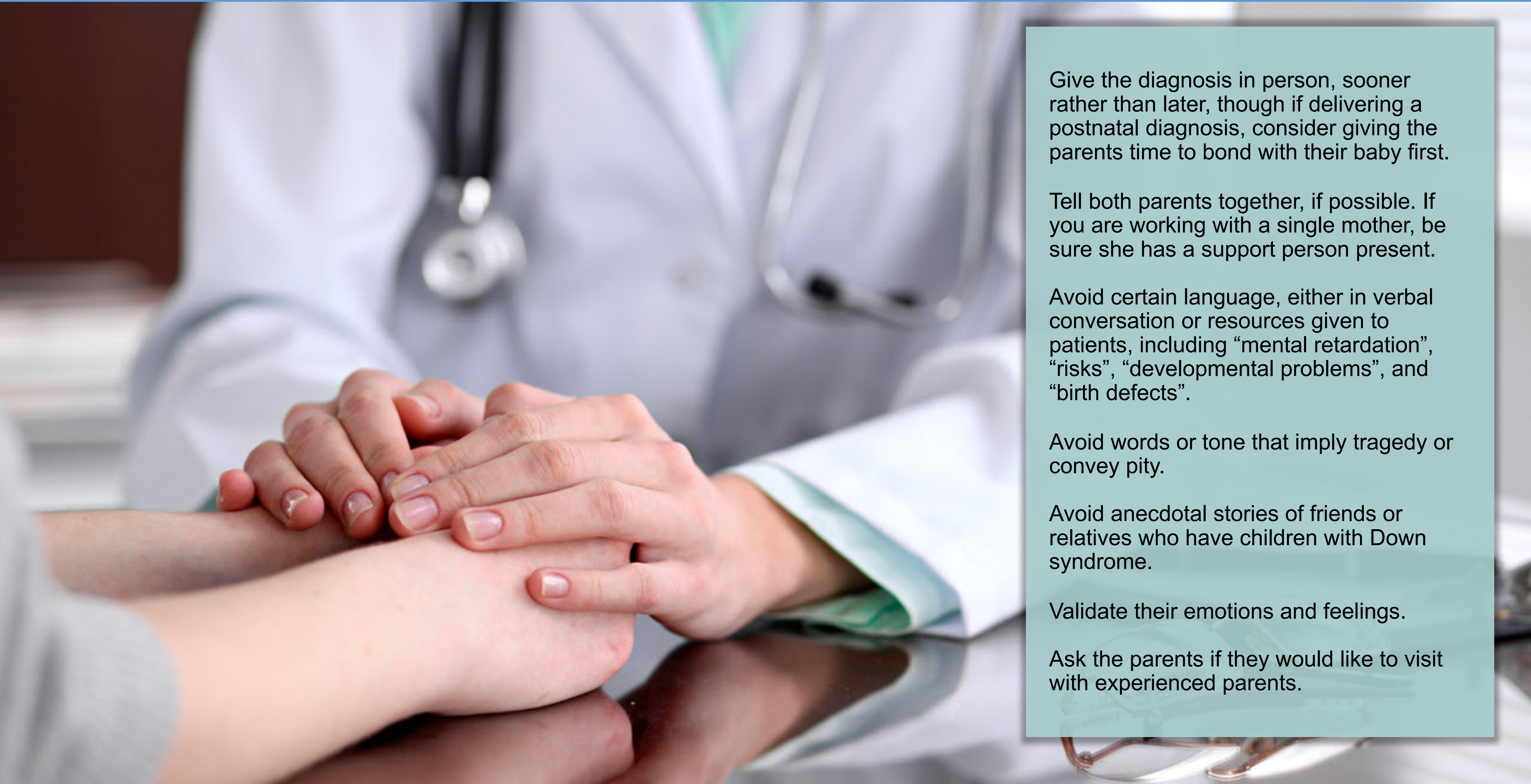
Presenting a Diagnosis



It cannot be stressed enough that everything you do and say when giving the diagnosis will have a profound impact on the parents.

It is something they will remember for the rest of their lives.

Presenting a Diagnosis



Give the diagnosis in person, sooner rather than later, though if delivering a postnatal diagnosis, consider giving the parents time to bond with their baby first.

Tell both parents together, if possible. If you are working with a single mother, be sure she has a support person present.

Avoid certain language, either in verbal conversation or resources given to patients, including “mental retardation”, “risks”, “developmental problems”, and “birth defects”.

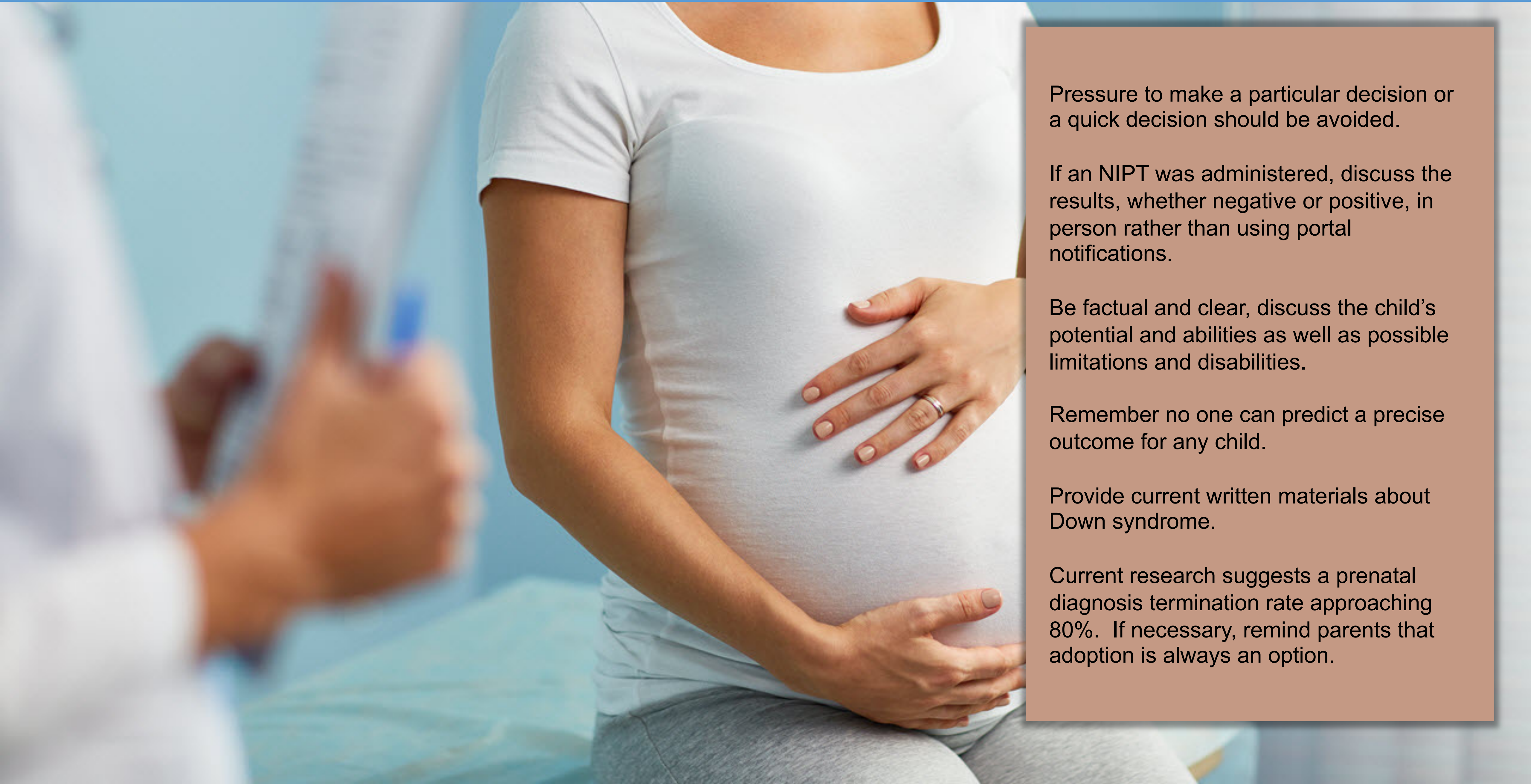
Avoid words or tone that imply tragedy or convey pity.

Avoid anecdotal stories of friends or relatives who have children with Down syndrome.

Validate their emotions and feelings.

Ask the parents if they would like to visit with experienced parents.

Delivering a Prenatal Diagnosis



Pressure to make a particular decision or a quick decision should be avoided.

If an NIPT was administered, discuss the results, whether negative or positive, in person rather than using portal notifications.

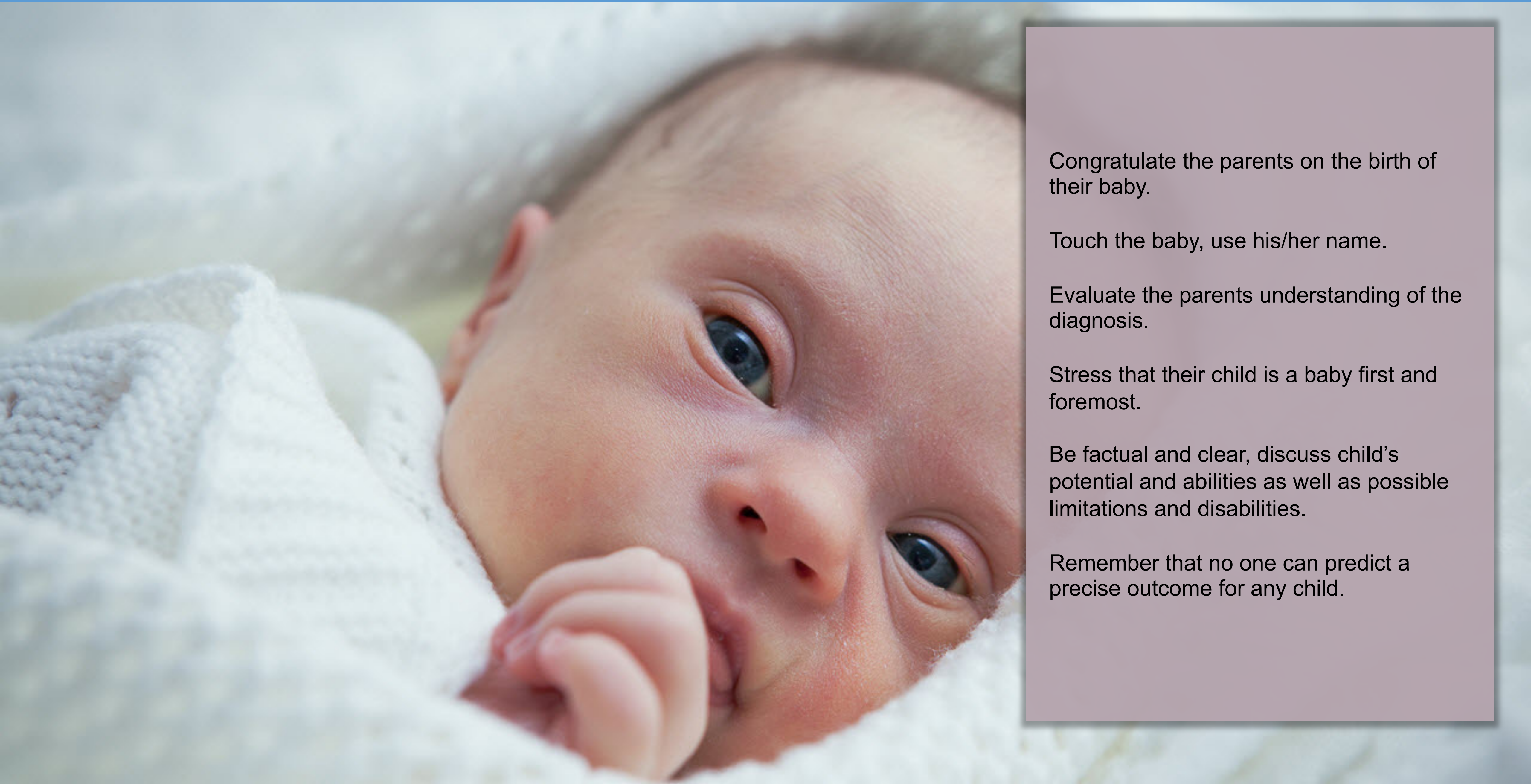
Be factual and clear, discuss the child's potential and abilities as well as possible limitations and disabilities.

Remember no one can predict a precise outcome for any child.

Provide current written materials about Down syndrome.

Current research suggests a prenatal diagnosis termination rate approaching 80%. If necessary, remind parents that adoption is always an option.

Delivering a Postnatal Diagnosis



Congratulate the parents on the birth of their baby.

Touch the baby, use his/her name.

Evaluate the parents understanding of the diagnosis.

Stress that their child is a baby first and foremost.

Be factual and clear, discuss child’s potential and abilities as well as possible limitations and disabilities.

Remember that no one can predict a precise outcome for any child.

Special Considerations

Be sure the entire team of health care professionals working with the new parents understands that consistent messaging is important.

Provide current and accurate information on Down syndrome – nothing more than 10 years old!

Make sure ALL health care professionals on the Labor/Delivery floor know where this information is housed.

It is extremely helpful for new parents to meet other parents of children who have Down syndrome.

How Will the Family Be Affected?

Brian Skotko, Co-director of Massachusetts General Hospital Down Syndrome Program led a research study in 2011.

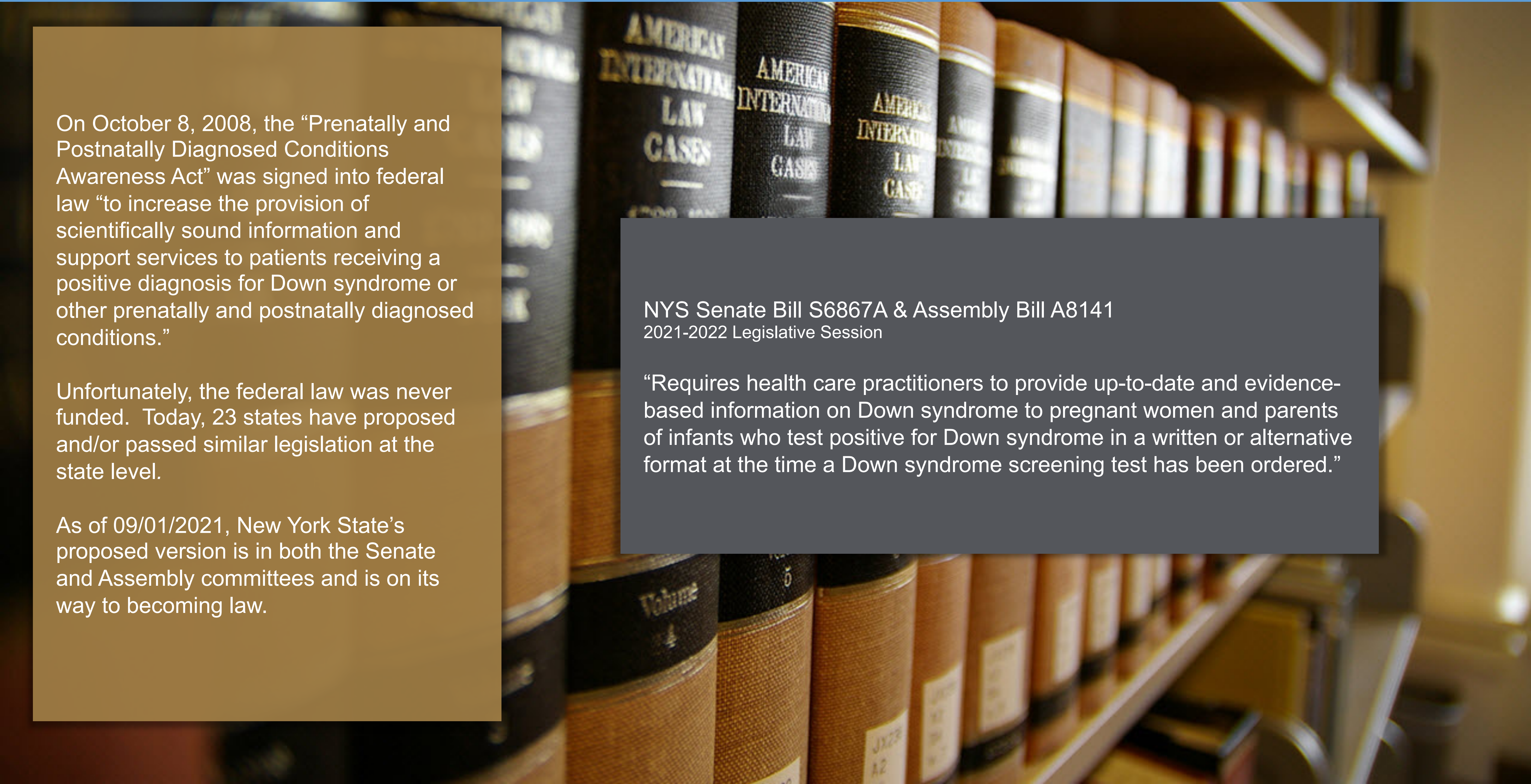
The study evaluated:

- 2000+ parents or guardians
- 800+ siblings
- 248 individuals with Down syndrome

Research found:

- 87% of families surveyed expressed overwhelming feelings of love and pride for family members with Down syndrome.
- Individuals with Down syndrome also reported feelings of happiness towards many areas of their lives.
- The extent to which individuals with Down syndrome were impaired had little or no relation to the attitudes within the family.

Where You Can Find Help – The Law



On October 8, 2008, the “Prenatally and Postnatally Diagnosed Conditions Awareness Act” was signed into federal law “to increase the provision of scientifically sound information and support services to patients receiving a positive diagnosis for Down syndrome or other prenatally and postnatally diagnosed conditions.”

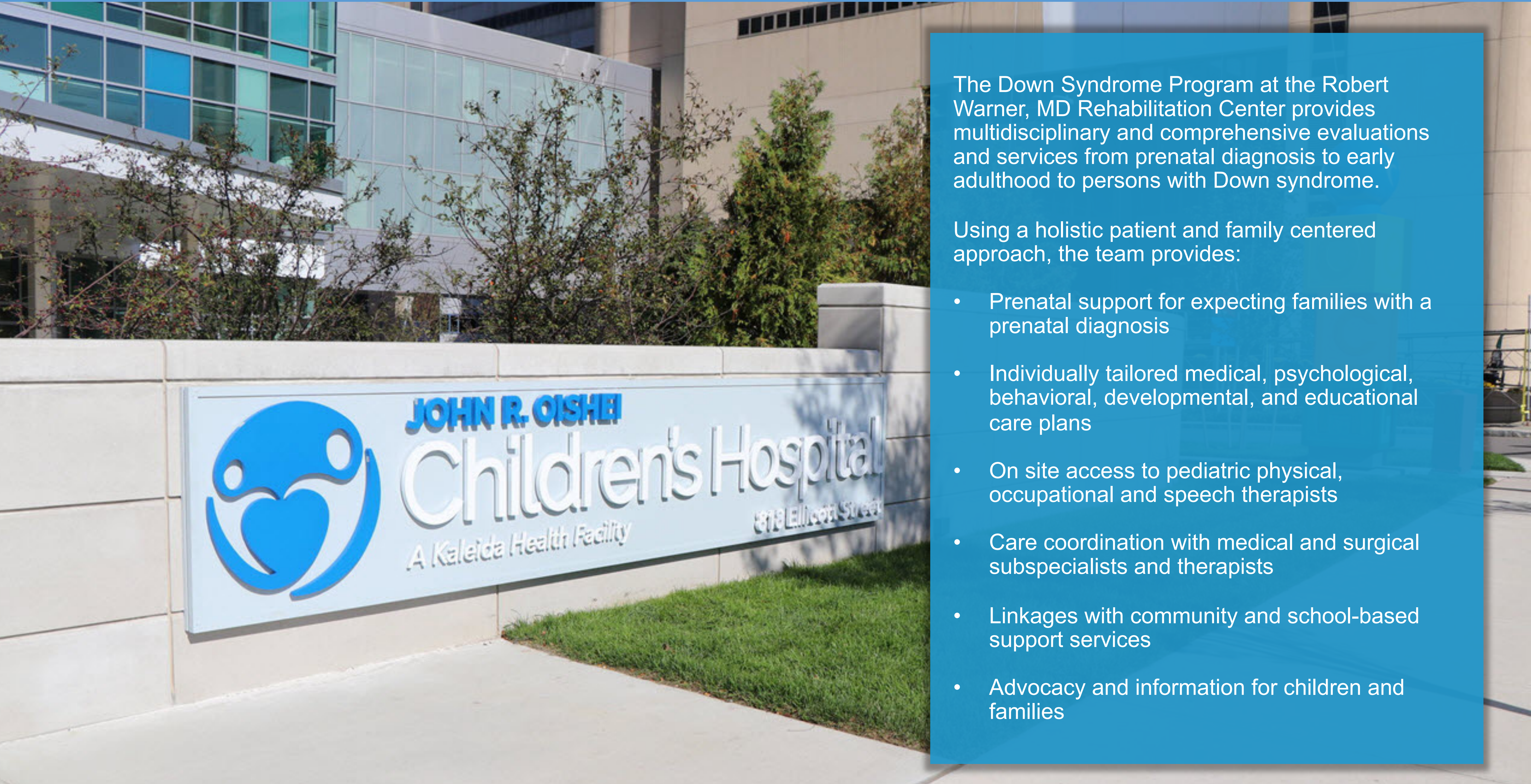
Unfortunately, the federal law was never funded. Today, 23 states have proposed and/or passed similar legislation at the state level.

As of 09/01/2021, New York State’s proposed version is in both the Senate and Assembly committees and is on its way to becoming law.

NYS Senate Bill S6867A & Assembly Bill A8141
2021-2022 Legislative Session

“Requires health care practitioners to provide up-to-date and evidence-based information on Down syndrome to pregnant women and parents of infants who test positive for Down syndrome in a written or alternative format at the time a Down syndrome screening test has been ordered.”

Where You Can Find Help – Medical Specialists



The Down Syndrome Program at the Robert Warner, MD Rehabilitation Center provides multidisciplinary and comprehensive evaluations and services from prenatal diagnosis to early adulthood to persons with Down syndrome.

Using a holistic patient and family centered approach, the team provides:

- Prenatal support for expecting families with a prenatal diagnosis
- Individually tailored medical, psychological, behavioral, developmental, and educational care plans
- On site access to pediatric physical, occupational and speech therapists
- Care coordination with medical and surgical subspecialists and therapists
- Linkages with community and school-based support services
- Advocacy and information for children and families

Where You Can Find Help – Community Programs

GiGi's Playhouse Buffalo
326 Kenmore Avenue
Buffalo, NY 14223

(716) 322-0010
buffalo@gigisplayhouse.org
gigisplayhouse.org/buffalo

Program Info & Schedule:
gigisplayhouse.org/buffalo/programs

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GiGi's Playhouse empowers families and provides free therapeutic, educational and career programs for children and families at all stages of development.

From prenatal diagnosis through career skills, families are never alone

Where You Can Find Help – The New Family Celebration Program

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families • self-advocates • friends
dspgwny

21 Connect

Family & Friends
DSA
Down Syndrome Association
Of Niagara, Inc.

- Presented by the WNY Down syndrome associations, the New Family Celebration Program welcomes new families into the Down syndrome community.
- Families are often reeling from the change in direction their lives have taken, and they cannot yet imagine the ways their little one will bless them. We meet them in the hospital, your office, or their home; say congratulations, give them support and guidance, and present them with a New Family Welcome Basket.
- The basket is designed to welcome new families into the community in a loving, caring, and fully inclusive way.
- The baskets contain many thoughtful items that any new or expanded families will need as well as items specific to the needs of babies born with Down syndrome, including:
    - swaddles and blankets
    - books and toys
    - onesies and bibs
    - highly regarded and reviewed informational/educational materials
    - information on local and national Down syndrome associations
    - Early Intervention and Medicaid support system guides
    - a special welcome letter

Where You Can Find Help – Local and National Associations

GiGi's Playhouse Down Syndrome Achievement Centers
716-322-0010
buffalo@gigisplayhouse.org
www.gigisplayhouse.org/buffalo

Down Syndrome Parents Group of WNY
716-832-9334
www.dspgwny.org

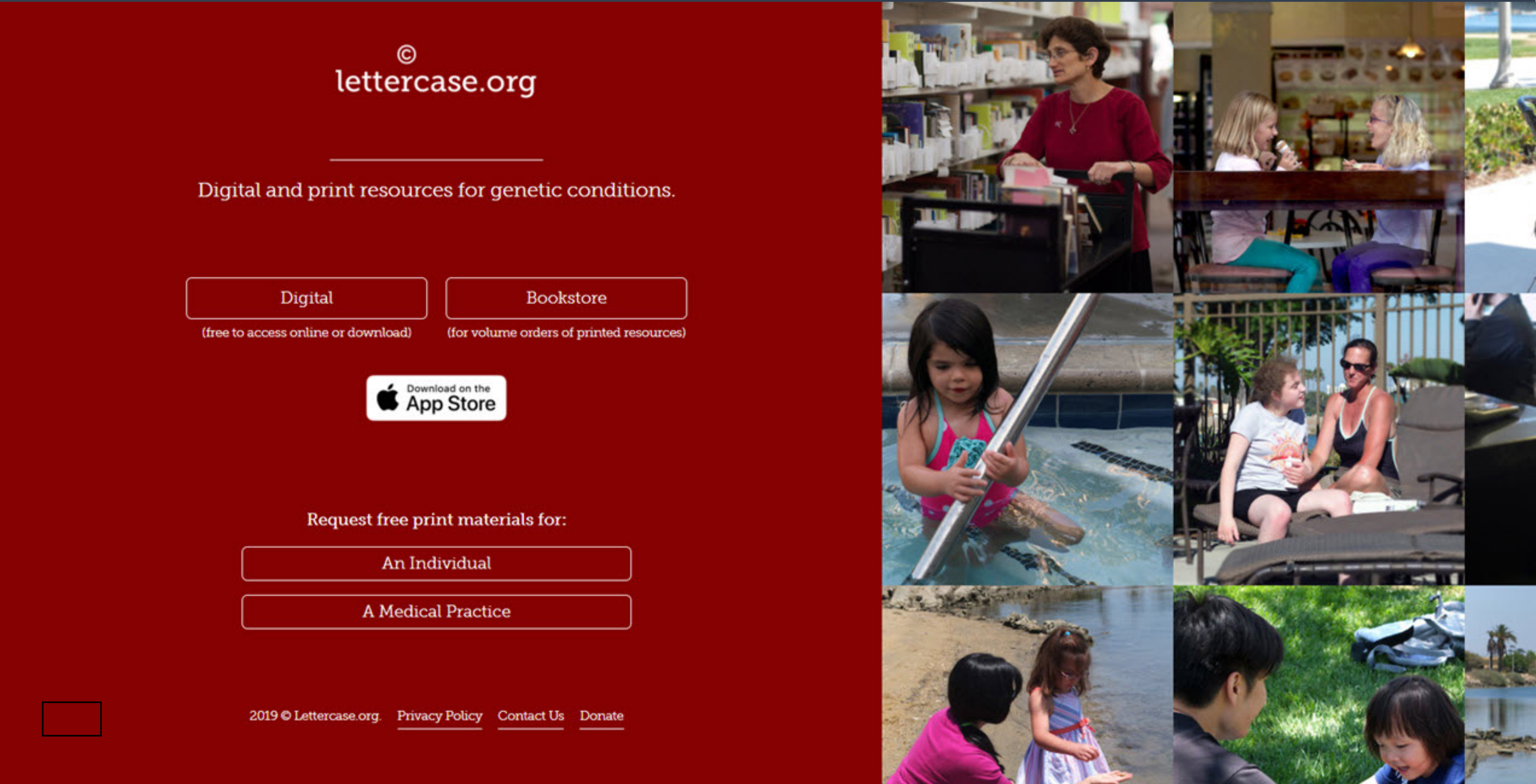
21_Connect
716-427-3575
connect@21connect.org
www.21connect.org

Parent Network of WNY
716-332-4170
info@parentnetworkwny.org
www.parentnetworkwny.org

National Down Syndrome Congress
800-232-6372
info@ndsccenter.org
www.ndsccenter.org

National Down Syndrome Society
800-221-4602
info@ndss.org
www.ndss.org

Where You Can Find Help– Current Resources



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Digital and print resources for genetic conditions.

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(free to access online or download)

Bookstore

(for volume orders of printed resources)

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Questions?

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Thank you