

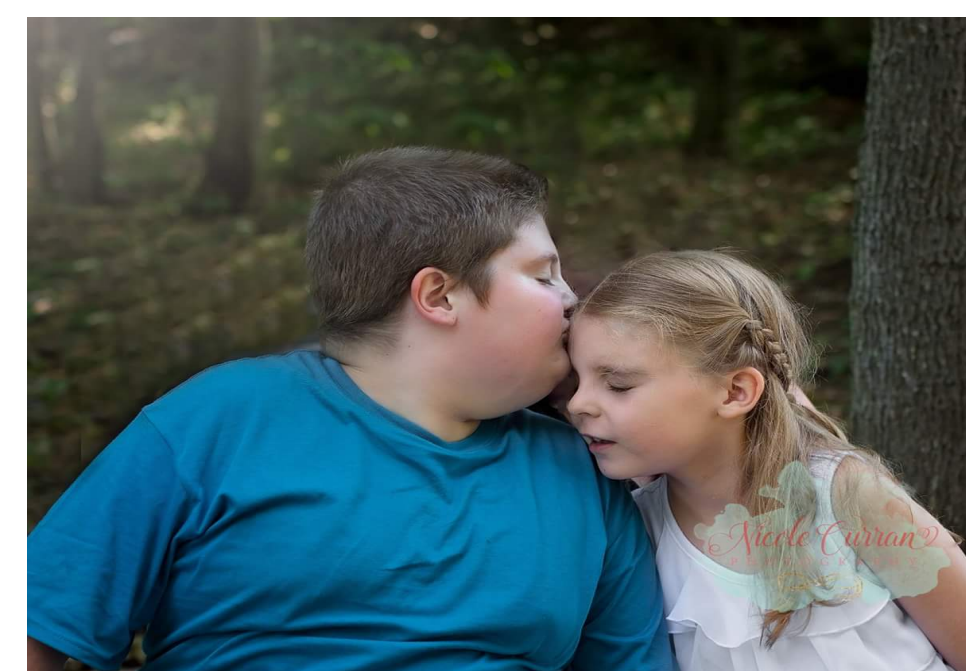


## Exploration in Genetic Disorders through Family Stories

### What is a Genetic Disorder?

A genetic disorder is a condition caused by a change in DNA ([www.medlineplus.gov](http://www.medlineplus.gov)). These disorders can be inherited or can occur randomly without a family history. Sometimes they are impacted by the environment.

- There are different types of genetic disorders ([www.medlineplus.gov](http://www.medlineplus.gov)):
  - Single-gene disorders such as Sickle cell anemia, where a mutation affects one gene.
  - Chromosomal disorders, such as Down Syndrome, where chromosomes are missing or changed.
  - There are also Complex disorders, such as certain cancers, where there are mutations in two or more genes.
- One difference between a genetic disorder and other conditions may be the emotional burden and stress related to passing the disorder on to their children. There is also a great concern for how it will affect future generations.
- Newborn screening is a public health service that tests for serious genetic conditions at birth ([www.babysfirsttest.org](http://www.babysfirsttest.org)). It is performed soon after the birth of the baby and allows doctors to be able to start treatment before any harmful effect occur. Newborn screening includes a hearing test and a simple blood test (a few drops). It can currently screen for more than 60 disorders.



### What is GEMSS? Why is it Important?

Genetics Education Materials for School Success (GEMSS) is a website that provides information to schools, families and healthcare providers about children with genetic disorders.

- It describes 37 genetic disorders and identifies supports that may help in school, with information that is vetted by NERGN experts and up to date.
- Perhaps most importantly, it tells the story of a child whose life is impacted by the condition, featured in the “Meet a Child” stories.

### My Activities

#### Family Stories

- Completed three practice interviews beginning with a friend, then with a mentor, then with a familiar family volunteer
- Completed interview and developed a story: Logan and MJ’s family - Where are they now?

#### Template for Interviews (Initial and Follow-up)

- Interviews took 60-90 minutes
- Prepared by researching condition and family story
- Created open-ended questions focusing on success



#### Video

- Created brief video highlighting my own experience throughout school and going to college
- Objective was to inspire others with communication differences to succeed

The family story and video will be available on the GEMSS website: [www.gemssforschools.org](http://www.gemssforschools.org)

### What I Learned

#### Importance of families to GEMSS

- Families are often the most significant relationships any child will have. They help children find resources that they need in order to be successful in their own way, happy and healthy.

#### Similarities in the Meet a Child stories

- Future generations may be impacted
- Parents and families face additional emotional burdens
- Families are often brought together and strengthened
- Siblings may develop significant empathy

#### Personal Growth

- Learned new technologies to support my own learning as an individual with communication differences
  - Hosting Zoom meetings
  - Recording conversations so I don’t miss information
- Learned more about the complexities of genetic disorders
- Developed leadership skills
  - Using my own story as the topic of an inspirational video
  - Coordinating meetings with families and mentor

The mission of the NH-ME LEND Program is to prepare leaders to support children with neurodevelopmental disabilities. Another program at The Institute on Disability / UNH is the New England Regional Genetics Network (NERGN). Their mission is to promote and improve health of people with inherited conditions. One of NERGN’s signature projects is the Genetics Education Materials for School Success (GEMSS) website, which supports children in schools. All three promote improvement to the lives of people with healthcare challenges.

[www.gemssforschools.org](http://www.gemssforschools.org)

