Children's Hospital of Philadelphia®
Roberts Individualized Medical Genetics Center
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Medical Intake Form

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Please complete this questionnaire as best you can. If you don't know the answer to a question please mark "?" next to the question.

Date: _____

Child's Name: _____

Date of Birth: _____ **Child's Age:** _____

Sex: Male ____Female _____

Parent/Guardian's name:
Father: _____ DOB: _____
Mother: _____ DOB: _____

Address: _____ **Phone:**
_____

_____

Email: _____

Ethnicity: _____

Where does your child live? Home ☐ Residential Facility: ☐ Both: ☐

Child's pediatrician: _____ **Phone #:**

Age at Diagnosis: _____ **Who made or suspected your child's diagnosis:**

What concerns do you have?

Pregnancy/Birth History:

Age at time of pregnancy: _____

Did you have ultrasounds completed during the pregnancy? Yes _____ No _____
If so, how many (approximately)? _____

Were any of the following findings detected by ultrasound?

- ☐ **Increased nuchal thickness** Yes ☐ No ☐
- ☐ **Shortening of long bones** Yes ☐ No ☐
- ☐ **IUGR (intrauterine growth retardation)** Yes ☐ No ☐
- ☐ **Micrognathia (small chin)** Yes ☐ No ☐
- ☐ **Microcephaly (small head circumference)** Yes ☐ No ☐
- ☐ **Large size** Yes ☐ No ☐
- ☐ **Diaphragmatic hernia** Yes ☐ No ☐
- ☐ **Kidney difference** Yes ☐ No ☐
- ☐ **Oligohydramnios** Yes ☐ No ☐
- ☐ **Polyhydramnios** Yes ☐ No ☐
- ☐ **Cardiac difference** Yes ☐ No ☐
- ☐ **Cleft palate** Yes ☐ No ☐
- ☐ **Ventriculomegaly (enlarged spaces in the brain)** Yes ☐ No ☐
- ☐ **Limb difference** Yes ☐ No ☐
- ☐ **Extra digits** Yes ☐ No ☐
- ☐ **Accessory nipples** Yes ☐ No ☐
- ☐ **Other differences (please list):**

_____
_____
_____

Did you have prenatal genetic testing? Yes ☐ No ☐
If so, what type?

_____Noninvasive prenatal genetic testing (NIPT) Results: _____
_____First Trimester Screening Results: _____
_____Sequential Screening Results: _____
_____Amniocentesis Results: _____
_____Chorionic villus sampling (CVS) Results: _____

Did you have a: vaginal delivery_____or C-section_____?
If C-section, what was the reason: _____

Birth Weight: _____ **Birth Length:** _____

Gestational age at delivery _____

Head Circumference _____

Apgar scores at 1 min _____5 min _____

After birth, did your child go to the NICU (neonatal intensive care unit)? Yes _____No _____
If yes, why? _____

Duration in Hospital after birth: _____

Medical History:

Gastroenterology

Does your child have any of the following:

- ☐ **Feeding Problems** Yes _____No _____
  If yes, what kind of feeding problems? _____
- ☐ **Dysphagia (difficulty swallowing)** Yes ☐ No ☐
- ☐ **Constipation** Yes ☐ No ☐
- ☐ **Pyloric Stenosis** Yes ☐ No ☐
- ☐ **Intestinal malrotation** Yes ☐ No ☐
- ☐ **Congenital diaphragmatic hernia** Yes ☐ No ☐
- ☐ **Gastroesophageal reflux (GERD)** Yes ☐ No ☐
  If yes, how was it treated? _____
  If yes, has reflux improved? _____
  If yes, has reflux resolved? _____

Has your child ever had:

pH probe	Yes _____ No _____	If yes, Results: _____
Milk scan	Yes _____ No _____	If yes, Results: _____
Upper GI	Yes _____ No _____	If yes, Results: _____
Endoscopy	Yes _____ No _____	If yes, Results: _____

Other: _____

Other GI problems: _____

Has your child ever had a fundoplication? Yes _____No _____
If yes, how old was he/she? _____

Does your child have a feeding device (NG tube, G-tube, etc.)
Yes _____No _____
If yes, what kind and at what age? _____

Does your child eat any food by mouth? Yes _____No _____
If yes, what foods/formula does your child eat? _____

At what age did your child first eat foods by mouth? _____

Has your child ever had any other abdominal surgery? Yes ☐ No ☐
If yes, what kind and why? _____

Pulmonology:

Does your child have a history of the following:

- ☐ **Aspirations** Yes _____ No _____
- ☐ **Recurrent upper respiratory infections** Yes _____ No _____
- ☐ **Pneumonia** Yes _____ No _____
- ☐ **Apnea** Yes _____ No _____
- ☐ **Asthma** Yes _____ No _____
- ☐ **Other respiratory problems:** _____

Was your child ever intubated? Yes _____ No _____ Age _____
Was any oxygen required? Yes _____ No _____ Age _____
Was this child on a ventilator? Yes _____ No _____ Age _____

Immunology:

Has your child ever have labwork completed to evaluated their immune system?
Yes _____ No _____

Did your child have a documented immunodeficiency? Yes _____ No _____
If yes, what is the treatment/management plan: _____

Cardiology:

Has your child ever had an echocardiogram? Yes _____ No _____

Does your child have a cardiac difference? Yes _____ No _____
If yes, what was the difference: _____
What has the treatment or management to date? _____
Does your child have any heart murmurs? Yes _____ No _____
What has been the treatment or management to date? : _____

Other cardiac concerns:

_____

_____

Genitourinary:

Has your child had a renal ultrasound? Yes _____ No _____

If yes, Results: _____

Does your child have any of the following kidney problems:

- ☐ **Ureteral reflux** Yes ☐ No ☐
- ☐ **Dysplastic kidney(s)** Yes ☐ No ☐
- ☐ **Single kidney** Yes ☐ No ☐
- ☐ **Small kidneys** Yes ☐ No ☐
- ☐ **Hematuria (blood in urine)** Yes ☐ No ☐
- ☐ **Proteinuria (protein in urine)** Yes ☐ No ☐
- ☐ **Other:** _____

Does your child have any of the following genital differences:

- ☐ **Undescended testes (males only)** Yes _____ No _____
- ☐ **Hypospadias (males only)** Yes _____ No _____
- ☐ **Other genitourinary concerns:**

_____

_____

Neurology:

Has your child had brain imaging studies (MRI, CT scan)? Yes _____ No _____

If yes, what studies were done?

_____

What were the results?

_____

Has your child had seizures? Yes ☐ No ☐

If so, what type of seizures? _____

When did they begin? How often do they occur? _____

What medications, if any, is he/she taking? _____

Has your child had an EEG study done? Yes ☐ No ☐

If yes, Results: _____

Has your child ever been diagnosed with the following?

- ☐ **Hypotonia (low muscle tone)** Yes ☐ No ☐
- ☐ **Hypertonia (increased muscle tone)** Yes ☐ No ☐
- ☐ **Tumor on the pineal gland** Yes ☐ No ☐
- ☐ **Degeneration/deterioration of the brain** Yes ☐ No ☐
- ☐ **Cerebral atrophy** Yes ☐ No ☐
- ☐ **ADHD** Yes ☐ No ☐
- ☐ **Autism** Yes ☐ No ☐

If yes please explain:

_____

_____

- **Regression (loss of skills)** Yes _____ No _____
  If yes please explain:

  _____

  _____

- **Other behavioral problems:** Yes _____ No _____
  If yes please explain:

  _____

  _____

Does your child sleep through the night? Yes ☐ No ☐

- **With Medication?** Yes ☐ No ☐

If yes, what medication? _____

Orthopedics:

Has your child ever had an x-ray of their forearm? Yes _____ No _____
If yes, Results: _____

Has your child ever had an x-ray of their hands? Yes _____ No _____
If yes, Results: _____

Has your child ever had any of the following limb differences:

- **Contractures** Yes ☐ No ☐
- **Radial-ulnar synostosis** Yes ☐ No ☐
- **Small feet** Yes ☐ No ☐
- **Syndactyly (fusion) of the 2nd and 3rd toes** Yes ☐ No ☐
- **Small hands/fingers** Yes ☐ No ☐
- **Missing forearm** Yes ☐ No ☐
- **Missing fingers** Yes ☐ No ☐

If yes, please describe any bracing, physical therapy or occupational therapy received: _____

_____

Has your child had any limb surgery? Yes ☐ No ☐
If yes, describe: _____

Otolaryngology (Ear, Nose, Throat):

Has your child had a hearing evaluation? Yes _____ No _____

- **If yes, was a hearing loss detected?** Yes _____ No _____
- **Does your child wear hearing aids?** Yes _____ No _____

Is the hearing loss:

- **Bilateral (both sides) _____ Unilateral (one side) _____**
- **Conductive _____ Sensorineural _____ Mixed _____**
- **Mild _____ Moderate _____ Severe _____ Profound _____**

Has your child's hearing loss:
Worsened _____ Improved _____ Remained Stable _____

Has your child had recurrent ear infections? Yes _____No _____

Has your child has tubes placed? Yes _____No _____
If so, how many times: _____

Does your child have a cleft palate? Yes _____ No _____ If yes, did it require repair? Yes _____ No _____ When?
Does your child have a cleft lip? Yes _____ No _____

Other otolaryngology concerns:

_____

_____

Endocrinology:

Has your child ever had hypoglycemia (low blood sugar)? Yes _____No _____
If yes, describe: _____

Has your child ever been tested for growth hormone deficiency? Yes _____No _____
If yes, results: _____

Has your child ever been on growth hormone therapy? Yes _____No _____
If yes, for how long: _____

Dental:

Does your child have any oral/dental problems? Yes _____No _____

Have you ever been told your child has cavities? Yes _____No _____

Do you think your child has cavities? Yes _____No _____

Have you ever been told your child has missing teeth? Yes _____No _____

Have you ever been told your child has crowded teeth? Yes _____No _____

Does your child have malformed teeth? Yes _____ No _____

Does your child have discolored teeth? Yes _____ No _____

Do you have trouble brushing your child's teeth? Yes _____ No _____

Has your child ever been seen by a dentist? Yes _____ No _____

Have you had difficulty finding a dentist who will treat your child? Yes _____ No _____

When did your child get his/her first baby tooth? _____

When did your child het his/her first permanent tooth? _____

Ophthalmology:

Does your child have any of the following:

- ☐ **Myopia (nearsighted)** Yes ☐ No ☐
- ☐ **Hyperopia (farsighted)** Yes ☐ No ☐
- ☐ **Nystagmus (involuntary rapid eye movement)** Yes ☐ No ☐
- ☐ **Strabismus (crossed eyes)** Yes ☐ No ☐
- ☐ **Tear (lacrimal) duct obstruction** Yes ☐ No ☐
- ☐ **Ptosis (drooping)** Yes ☐ No ☐
- ☐ **Glaucoma** Yes ☐ No ☐
- ☐ **Cataracts** Yes ☐ No ☐
- ☐ **Other:** _____

Has your child had ophthalmologic surgery? Yes ☐ No ☐

If yes, describe: _____

Other ophthalmologic concerns:

_____

_____

Dermatology:

Does your child have any of the following:

- ☐ **Eczema** Yes ☐ No ☐
- ☐ **Hemangiomas** Yes ☐ No ☐
- ☐ **Skin discoloration** Yes ☐ No ☐
- ☐ **Birthmarks** Yes ☐ No ☐
- ☐ **Other**: _____

Development:

Has your child reached the following milestones:

- ☐ **Rolled** Yes ☐ No ☐ At what age:_____
- ☐ **Sat** Yes ☐ No ☐ At what age:_____
- ☐ **Crawled** Yes ☐ No ☐ At what age:_____
- ☐ **Walked** Yes ☐ No ☐ At what age:
- ☐ **Talked** Yes ☐ No ☐ At what age:_____
- ☐ **Toilet Trained** Yes ☐ No ☐ At what age:_____
- ☐ **Dress Self** Yes ☐ No ☐ At what age: _____

Briefly summarize your child's current developmental abilities:

What kind of school program (if applicable) is your child enrolled in?

Does your child have any:

Self-injurious behaviors Yes _____No _____

If yes, describe: _____

Aggressive behaviors Yes _____No _____

If yes, describe: _____

Please briefly describe your child's current developmental abilities, as well as any therapies your child is receiving:

Genetics:

Has your child had any of the following genetic studies:

- ☐ **Chromosome/Karyotype** Yes _____No _____ Result: _____
- ☐ **Microarray** Yes _____No _____ Result: _____
- ☐ **AFF4** Yes _____No _____ Result: _____
- ☐ **Exome** Yes _____No _____ Result: _____
- ☐ **Genome** Yes _____No _____ Result
- ☐ **Other genetic studies and results:**

_____

Has your child enrolled in Dr. Krantz's Research study at CHOP? Yes _____No _____
If no, are you interested in learning more about research opportunities at CHOP? Yes _____No _____

Medications:
Please list all the medications that your child is presently on or has been on in the past.

Medication	Dosage	Start date/Age	Stop date/Age	Reason for medication

Surgeries and Procedures:
Please list any surgeries or procedures your child has had or is scheduled to have.

Month/Year	Procedure