How to use this guide:

1. *Print this guide double-sided*
2. *Fold the pages in half along the dotted line to form a booklet*
3. *Fill out this guide to help you prepare for your appointment*
4. *Bring the guide to your genetic counseling appointment to use as a reference during the discussion*

Ambit CARE

Genetic Counseling Discussion Guide

Please note: With rare diseases, it is important to discuss all of your child's symptoms with a healthcare professional. AmbitCARE provides this guide for the benefit of the rare disease community, but AmbitCARE is not a medical provider, nor a health care facility and, thus, can neither diagnose any disease or disorder nor endorse or recommend any specific medical treatments. This document should be used along with other resources as a guide to help prepare for your child's genetic counseling appointment. This guide is not a substitute for seeking the personal and individualized medical advice of a qualified health care professional regarding your child's particular diagnosis, cure, or treatment of a condition or disorder.

Learn more at AmbitCare.com

Make the most of your child's genetic counseling appointment by showing up prepared. With the most accurate clinical picture of your child, genetic counselors and genetic physicians can make the best recommendation about what genetic test(s) are most relevant. No symptom is too small to address. Do not be afraid to ask questions and seek further explanation. This guide is meant to help you prepare for your child's appointment but is not intended to be a substitute for seeking medical care from a trained professional.

BASIC PATIENT INFORMATION

Patient Name: _____ **DOB:** ____ / ____ / ____

Age: _____ **Sex Assigned at Birth:** ☐ Male ☐ Female ☐ Intersex

Height: _____ **Weight:** _____ **Head Circumference:** _____

Ethnicity:

☐ American Indian or Alaska Native ☐ Asian ☐ Black or African American ☐ White
☐ Hispanic or Latino ☐ Native Hawaiian or Other Pacific Islander ☐ Other

Why are you pursuing genetic testing for the patient?

☐ Concern of a genetic condition
☐ The patient's PCP recommended genetic testing
☐ Physicians have not been able to explain the cause of the patient's symptoms
☐ I would like to know risk information for my family members
☐ Other (describe) _____

The following questions are only relevant if your child's PCP recommended genetic testing. If your child's PCP did not recommend genetic testing, skip to next section.

Why did the patient's PCP recommend genetic testing?* _____

At what age did you or your child's physician suspect something was wrong? _____

What caused you or your physician to suspect something was wrong? _____

**If you do not know why the patient's PCP recommended genetic testing, seek out that information from the PCP's office.*

OFFICIAL DIAGNOSES

Which of the following has your child been diagnosed with? *Check all that apply.*

☐ **Autism Spectrum Disorder:** Age at diagnosis: ____ Diagnosing Physician: _____
☐ **Developmental Delay:** Number of months the patient is behind in development: ____
☐ **Intellectual Disability:** IQ: _____
☐ **Epilepsy / Seizures:** What age was the patient at first seizure?: _____
☐ **2 or more birth defects requiring surgery**
☐ **Cerebral Palsy:** Age at diagnosis: ____ Diagnosing Physician: _____
☐ **Other Diagnoses:** _____

MEDICATIONS

Is your child currently on any medications? ☐ Yes ☐ No

If you answered yes, please specify:

Is your child currently taking any vitamins or supplements? ☐ Yes ☐ No

If you answered yes, please specify:

EARLY INTERVENTION & THERAPY

Did your child qualify for any services after an early intervention evaluation? ☐ Yes ☐ No

If you answered yes, please specify:

Indicate what therapies your child has received and when they received each therapy (in years). *If the child is still receiving the therapy, write "TODAY" as the end date.*

☐ **Physical Therapy** Start & End Date: _____ to _____
☐ **Occupational Therapy** Start & End Date: _____ to _____
☐ **Speech Therapy** Start & End Date: _____ to _____
☐ **Behavioral Therapy** Start & End Date: _____ to _____
☐ **Other Therapies:**
_____ Start & End Date: _____ to _____
_____ Start & End Date: _____ to _____
_____ Start & End Date: _____ to _____

FAMILY HISTORY

Mother's Ethnicity:

☐ American Indian or Alaska Native ☐ Asian ☐ Black or African American ☐ White
☐ Hispanic or Latino ☐ Native Hawaiian or Other Pacific Islander ☐ Other

Father's Ethnicity:

☐ American Indian or Alaska Native ☐ Asian ☐ Black or African American ☐ White
☐ Hispanic or Latino ☐ Native Hawaiian or Other Pacific Islander ☐ Other

Are the patient's parents related to one another by blood? ☐ Yes ☐ No

How many siblings does the patient have? _____

For each sibling, list the gender, age, and whether the child is a full sibling, half sibling through mother, or half sibling through father.

Did the child's parents receive any carrier testing? ☐ Yes ☐ No *If yes, please specify:*

Has anyone in your family had a genetic evaluation or genetic test? ☐ Yes ☐ No

If you answered yes, please specify your child's relation to that family member, what that family member was tested for, and what the results were:

Ask those family members if they would be willing to send their genetic test results to your genetic counselor.

ORGAN BY ORGAN REVIEW

Please describe if your child has any symptoms or concerns in the following areas:

Heart:

Brain:

Kidneys:

Gastrointestinal Tract *(ex. birth defect of the stomach or intestines, constipation, or vomiting)*:

Lungs:

Skeletal System:

Skin:

Vision, Sight, or Hearing:

Muscle Tone:

Feeding *(ex. sucking, chewing, swallowing, foods that the child avoids or makes them sick)*:

Breathing:

Sleeping:

Behavior:

Are there any other abnormalities?

PATIENT HISTORY

At what age did your child reach the following milestones?

Roll Over: _____ Eye Contact: _____ Sit Independently: _____

Crawl: _____ Walk Independently: _____ Feed Themself: _____

Use Utensils: _____ First Words: _____ Use Phrases: _____

Use Sentences: _____

Were/Are there any notable delays?

_____

_____

Has your child regressed in any skills? ☐ Yes ☐ No *If yes, please specify:*

_____

Has your child ever experienced seizures? ☐ Yes ☐ No *If no, skip to the next section*

When did your child first start having seizures? _____

How often does your child have seizures? _____

What type of seizures has your child had? _____

Have you noticed any seizure triggers (ex. fever, hot baths, etc)? _____

BIRTH HISTORY

How many pregnancies did the patient's mother have before having the patient? _____

Number of miscarriages (if any): _____ Number of terminations (if any): _____

List any complications during the patient's pregnancy (illnesses, fever, bleeding)?

_____

What was the patient's gestational age at birth (estimate)? _____

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

Did the patient experience any prenatal exposures to alcohol, cigarettes, illicit drugs, or medications? ☐ Yes ☐ No *If yes, list below:*

_____

List any birth defects the patient was born with (whether surgically corrected or not).

_____

_____

TESTING

Has your child ever received genetic testing? ☐ Yes ☐ No ☐ I don't know

If you answered yes:

What genetic test did your child receive? _____

Describe any notable findings: _____

Did your child receive prenatal genetic testing? ☐ Yes ☐ No

If you answered yes:

Describe any notable findings: _____

Check the box if your child has received any of the following tests:

☐ **EEG**
Results: _____

☐ **MRI:**
Results: _____

☐ **CT Scan**
Results: _____

☐ **EKG**
Results: _____

☐ **Echocardiogram**
Results: _____

☐ **Ultrasound**
Results: _____

☐ **Metabolic Tests (blood or urine)**
Results: _____

☐ **Early Intervention Evaluation**
Results: _____

☐ **Neuropsych Evaluation**
Results: _____

☐ Other Tests: _____
Results: _____

HOSPITALIZATIONS

Please list the date and reason for any and all hospitalizations or emergency room visits in the last 2–5 years:

Date:	Reason for Hospitalization / ER Visit:	Notes: