

Genetics Referral Guide



The purpose of the Genetics Referral Guide is to help health care providers:

- recognize indications for referral of children to a genetics center for evaluation
- have easy access to contact information for regional genetics centers in the State of Ohio

When Making a Genetics Referral

The service coordinator, WIC staff, nurse, social worker, etc., should:

1. Request a referral from the child's primary care provider. Write letter to the physician regarding your concerns. Have family discuss the option with their primary care physician.
2. If the primary care provider will not grant a referral, the family or provider may call to schedule an appointment. (NOTE: Insurance may require a physician referral.)
3. In the referral, include a note to the geneticist with your concerns.

The primary care provider should:

1. Discuss the referral with the family.
2. If unsure that a genetics evaluation is necessary, discuss with a genetic counselor or geneticist in your region (see back).

The role of the health care provider is to communicate concerns in a sensitive manner and help the patient/family:

- Consider when a child's condition may be genetic.
- Understand the importance of a genetics evaluation.
- Schedule a genetics appointment.

The role of the geneticist/genetic counselor is to help the patient/family:

- Understand the diagnosis and medical facts.
- Determine if there is an underlying genetic or environmental cause.
- Clarify the recurrence risks.
- Access appropriate testing, clinical trials and support organizations.
- Know how to prepare for pregnancy and increase the chances for having a healthy baby.

Addressing concerns about cost/distance/time:

- Genetic evaluations and some genetic testing are covered by Medicaid and most insurance.
- Ohio's Bureau for Children with Medical Handicaps (BCMh) program may cover diagnostic and treatment related to the child's genetic condition.
- Genetics outreach clinic sites are available throughout the state.
- The genetics evaluation is usually completed in 1 or 2 appointments. Some children will have an annual follow-up.

Adapted with permission from the "Genetics Education for Early Onset Michigan Providers" training curriculum.

A child should have a genetic evaluation and/or counseling if they have:

Suggestions for the health care provider in asking questions

A genetic condition

1. Does your child have a known genetic condition?
2. I'm not familiar with your child's condition. Have you ever been told whether your child's condition is genetic?

Unexplained hearing loss

1. Are you concerned about your child's hearing?
2. Has your child ever failed a hearing screening or been diagnosed with hearing loss?
3. Have you ever been given a reason for your child's hearing loss?

Note: temporary, mild hearing loss related to ear infection is not considered a reason for referral to genetics. If you suspect hearing loss, refer for an audiology evaluation first.

Significant developmental delay/mental retardation

Note: if you suspect developmental delay refer for evaluation by a psychologist, OT/PT, developmental pediatrician and/or the local Help Me Grow program in the family's county of residence.

Unusual, progressive and/or regressive behavioral features

Note: be careful. It may be difficult to distinguish behavioral problems with a genetic etiology from other factors. In these cases, screen through child psychology, behavioral specialist or developmental pediatrician before considering a genetics referral.

1. Have you noticed unusual behaviors in your child? (Examples that may be characteristic of a syndrome include: self-injury, hand-wringing, arm-flapping, walking up the legs with hands, toe walking, inappropriate laughing, unusual food-seeking or food-avoiding behaviors, repetitive behaviors, etc.)
2. Has your child been officially diagnosed with autism by an autism specialist?
3. Has the child's behavior been getting worse with time?
4. Have you noticed any loss of skills in your child?
Examples:
a. Child loses ability to crawl, walk, sit alone, hold head erect, etc.
b. Child with about 20 words or so becomes nonverbal
c. Child with normal pincer grasp loses ability to grasp toys and instead continuously keeps hands in mouth

Neurological abnormalities

1. Does the child have seizures, abnormal movements, low muscle tone, spasticity (increase over the normal tone of muscle), tremors, etc.?

Note: if yes, a referral to genetics should be secondary to an evaluation by neurology.

Specific medical conditions

1. Has the child been given any medical diagnoses that we have not talked about? Examples; congenital heart disease, kidney disease, etc.

Multiple affected relatives with the same condition

1. Does anybody else in the family have the same diagnosis as your son or daughter?
2. Is there anybody else in the family with a child like your son or daughter?
3. Are there any traits running in the family (such as extra fingers, white patch of hair)?

A child should have a genetic evaluation and/or counseling if they have:

Suggestions for the health care provider in asking questions

Major birth defects

Note: Some major birth defects are obvious and will be openly shared with you. Others may be internal or already surgically corrected.

1. Was the baby born with any birth defects? (e.g. club feet, cleft lip, heart defects)
2. Did the child require any surgeries in the newborn period? (often related to birth defects)

Two or more minor birth defects (dysmorphic features)

1. Was the baby born with any unusual physical features? (Examples: webbing between the fingers or toes, umbilical hernias, ear tags, undescended testes, droopy eyelid, etc.)

Note: care must be taken when addressing dysmorphic features in children. A child with multiple dysmorphic features often does not resemble the parents or may resemble a parent who also looks "unusual."

1. Who do you think the child resembles in the family?
2. Do you notice any features in the child that seem different than other family members?
3. When you look at your family photos, whom does the child look like?

Note: if you have identified dysmorphic features (see dysmorphology checklist) in the child and feel a genetic evaluation would be beneficial, you may be the first to approach the family about these features. Choose your words carefully. You can contact a genetic counselor if you are unsure how to approach a family regarding dysmorphic features.

Medical problems and their parents are closely related blood relatives

Note: just because a child has related parents does not mean they will automatically experience genetic problems. The term "close relatives" is usually defined as second cousins or closer.

1. Is there any possibility you and your husband/wife are related by blood?
2. Is your family related to your husband/wife's family? For example, are you cousins?
3. Do you and your husband/wife have a common ancestor (e.g. grandparent)?

Note: remember that marrying relatives is accepted and encouraged in some cultures. Be sure to reassure the family that you ask all couples this question as a screen for genetic conditions.

Abnormal growth, not related to prematurity

1. Has the doctor had any concerns about your child's growth?
2. Has the pediatrician mentioned that your child is unusually small for his/her age? Have they ever used the term "Failure to thrive" to describe your child?
3. Have any of the child's measurements been above or below the normal growth curve? For instance, does the child's head measure unusually large or small?
4. Have you ever noticed that one side of the child's body is bigger than the other? For instance, does one leg appear longer than the other?

Significant vision loss

Note: most children with mild visual problems requiring glasses do not need a genetic referral—screen through ophthalmology first. Also, vision problems as a result of retinopathy of prematurity are not genetic.

1. Do you have concerns about your baby's eyesight or have you ever been told your child has a vision problem?
2. Has your child ever had vision screening or any abnormal vision tests results?
3. Do you think your child can see appropriately?

Dysmorphology checklist

Minor malformations may not have substantial medical or cosmetic consequences. They can be normal variants within the general population. However, when children have two or more minor malformations (in one or more of the categories below), a genetic syndrome may be questioned. **This list is to be used as a guide and is not all inclusive. Unusual features should not be ignored. Care should be taken to incorporate the child's ethnic background.**

Head/skull— Abnormally large or small head; abnormal shape to head; backward sloping forehead; prominent or protruding forehead; early closure of the soft spots.

Face— Facial asymmetry (one side of face is larger than the other; eyes and/or ears appear uneven); premature aging; immature facial appearance; coarsening (lacking refinement or delicacy).

Ears— Large ears; simple ears; asymmetric ear size (ear length differs by more than 3mm); protuberant (projecting) ears; ear tags; ear pits; creases in ear lobe; abnormal shape (cupped, crumpled); abnormal placement (low-set, rotated toward the back of the head).

Eyes— Asymmetry of the eyes; protruding eyes; eyes very far apart; eyes very close together; abnormally formed eyelids; small eye openings; tumors of the eye; different colored eyes; the pupil (black opening at the center of the eye) is white; abnormal eye movement; eyes do not look straight ahead; droopy eyelid;

lens dislocation; eye openings that slant up or down; iris coloboma (absence or defect in the color part of the eye); the white part of the eye is blueish or gray; epicanthal folds (skin folds at the inner corner of the eye); eyebrows that have grown together.

Nose— Nasal pits (pits anywhere on the nose... may have hair growing); prominent, bulbous nose; broad nasal bridge.

Mouth— Abnormal tooth enamel; lower lip pits; abnormally colored teeth; large or prominent lips; long/flat philtrum (skin between upper lip and nose); wide-spaced teeth; abnormally shaped teeth (peg shaped, conical); missing numerous teeth; high-arched palate; thickened tongue; missing or split uvula (the structure at the back of the throat); poor coordination for eating; abnormally large or small mouth opening.

Chin— Large or small chin; chin placed too far back.

Neck— Redundant neck skin; torticollis (head constantly held to one side); short neck; pits or clefts in neck.

Body/chest— Absence of the clavicles (collar bones); absence of the chest muscles; extra or missing nipples; shield-shaped chest with wide spaced nipples; pigeon chest (chest sticks out); funnel chest (chest dips inward).

Abdomen— Absence of abdominal muscles; enlarged liver or spleen; umbilical hernia (stomach appears to stick out near the belly button due to pressure from the intestines).

Genitalia— Testicular hypoplasia (incomplete development); abnormally large testicles; micropenis; shawl scrotum (scrotum comes up around the penis).

Anus— Abnormally placed anus.

Back— Scoliosis; short trunk; “winged” shoulder blades; dimple in lower back with or without tuft of hair.

Skin— Abnormal skin creases; abnormal skin fragility; hyperelasticity (excessive stretching); absence of sweating glands; hypopigmentation (very light skin or spots of light skin); hyperpigmentation (dark spots); skin tumors; port wine stains; unusual birthmarks; albinism (lack of color in the skin, hair and eyes); scaling skin.

Hair— Sparse hair; early graying; white streak of hair (white forelock); low posterior (back part) hairline; hair over the midline spine; patchy hair; straight or droopy eyelashes; hair fragility; upsweep (hair tends to stick up naturally instead of laying flat) of hair; widow's peak; continuous eyebrow; excessive hair on the body; coarse hair.

Nails— Small nails; extremely slow-growing nails; absence of nails.

Limbs— Diminished muscle mass; bulky firm muscle; short limbs; bone fragility; disproportionate long limbs; limb asymmetry.

Joints— Enlargement; loose joints; multiple joint dislocations; limited joint movement; webbing of skin over joints.

Hands/feet— Short fingers/toes; small hands/feet; abnormally placed thumb; webbing; bent fingers; tapered (a gradual decrease in width/thickness) fingers; long fingers/toes; single palmar crease (one long crease in the center of the hand); broad thumbs/toes.

Behavioral/neurological— Low or high muscle tone; expressionless face; abnormal movements; ataxia (jerky movements); seizures; hyperactivity (unusually and constantly energetic and difficult to focus); hypoactivity (lack of energy or interest); inappropriate laughter; hand-wringing; self-abuse; abnormal gait.

Voice— High-pitched cry; hoarse voice; no voice.

Growth— Tall stature; short stature; disproportionate growth (trunk or limbs).

Other— Congenital hip dislocation; multiple fractures; unusual odors (particularly when ill), unusual dietary pattern (ex: avoids protein).

The Ohio Regional Comprehensive Genetics Services Network

REGION I

1 Division of Human Genetics
Cincinnati Children's Hospital Medical Center
3333 Burnet Avenue
Cincinnati, OH 45229
Phone: (513) 636-4760
www.cincinnatichildrens.org
Cincinnati Outreach Sites:
Hillsboro | Georgetown | Seaman

REGION II

2 Department of Medical Genetics and Birth Defects
Dayton Children's Hospital
One Children's Plaza
Dayton, OH 45404
Phone: (937) 641-3800
www.childrensdayton.org
Dayton Outreach Site: Beavercreek

REGION III

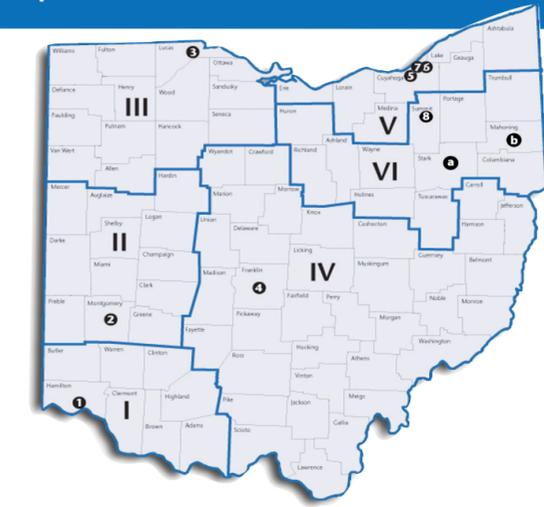
3 Northwest Associate Genetics Center
Mercy St. Vincent Medical Center
2222 Cherry Street-Suite 2300
Toledo, OH 43608
Phone: (419) 251-8012 or 1-877-322-2200
www.utoledo.edu
Toledo Outreach Sites:
Defiance | Lima | Bowling Green

REGION IV

4 Regional Genetics Center
Nationwide Children's Hospital
700 Children's Drive
Columbus, OH 43205
Phone: (614) 722-3535
www.nationwidechildrens.org
Columbus Outreach Sites:
Athens | Waverly | Marietta | Zanesville | Lima

REGION V

5 Regional Genetics Center
Department of Pediatrics
MetroHealth Medical Center
2500 MetroHealth Drive
Cleveland, OH 44109
Phone: (216) 778-4323
www.metrohealth.org/Clinical/Pediatrics



REGION VI

6 Center for Human Genetics
Case Western Reserve University and
University Hospitals Case Medical Center
11100 Euclid Avenue, Lakeside 1500
Cleveland, OH 44106
Phone: (216) 844-3936
www.uhhospitals.org

REGION VII

7 Center for Personalized Genetic Healthcare
Cleveland Clinic Foundation
9500 Euclid Avenue
Cleveland, OH 44195
Phone: (216) 636-1768 or
(800) 988-5686 (toll free)
www.clevelandclinic.org/childrens-hospital/specialties-services/what-we-treat/medical-genetics.aspx

REGION VIII

8 Regional Genetics Center
Children's Hospital Medical Center-Akron
One Perkins Square
Akron, OH 44308
Phone: (330) 543-8792
www.akronchildrens.org
Akron Outreach Sites:
Ashland | Beavercreek | Ravenna | Norwalk

The Ohio Department of Health funds seven Regional Comprehensive Genetics Centers (RCGC) in the State of Ohio. In addition to the seven main genetics centers, each RCGC also has outreach clinics in neighboring counties to increase access to genetic services. To make an appointment, contact the RCGC in your region.