

There can be several benefits of having a genetic diagnosis and apply to all conditions below.

- Timely access to services and supports through early intervention and appropriate educational programming
- Better understanding of genetic risk for family members or future pregnancies
- Connection with condition-specific support groups and resources.

Genetic condition that may be identified on the Essential Test <i>(in no particular order)</i>	First Signs of the condition may include the following	Healthcare recommendations that come with identification of the genetic diagnosis
16p12.2 deletion syndrome	Developmental delay, intellectual disability (ranging from mild to profound), autism spectrum disorder	Evaluations for possible heart malformations, seizures, psychiatric and behavioral problems, and/or hearing loss
Prader-Willi syndrome	Significantly low muscle tone (feeling limp or floppy) and feeding difficulties	Consideration of growth hormone therapy; monitor for neuropsychiatric conditions and extreme overeating
Williams syndrome	Low birth weight, feeding difficulties, high levels of calcium in the blood, low muscle tone, overly flexible joints, developmental delays	Evaluation for psychological and psychiatric problems (such as ADHD and anxiety), cardiology evaluation (heart issues seen in 75% of those with Williams syndrome), treatment of high calcium in blood which can cause seizures, possible hormone treatment for precocious puberty
Williams syndrome region duplication syndrome (also called 7q11.23 duplication syndrome)	Severe impairment in expressive language, developmental delays	Monitor for behavioral concerns such as anxiety, ADHD, oppositional defiant disorder, and ASD; surveillance for seizures (seen in 15-20% of those with the duplication); routine follow up as needed for birth defects such as heart defects, palate (roof of the mouth) problems, and kidney malformations
Distal 18q deletion syndrome	Hearing loss due to problems with ear canal development, microcephaly (small head size), developmental delays	ENT/Audiology evaluation for possible hearing loss and ear canal issues
22q11.2 deletion (also called DiGeorge syndrome)	Heart defect, palate abnormalities, developmental delays or learning difficulties, increased infections due to a weak immune system	A multidisciplinary clinic team approach can be tailored to the child's specific needs, prompt treatment of infections; growth hormone treatment as needed; surveillance and early intervention for psychiatric illnesses such as anxiety, ADD, and schizophrenia; evaluation for heart issues (seen in over 75% of individuals with the deletion)
47,XXY (also called Klinefelter syndrome)	Mild developmental delay, learning difficulties, attention disorders	Evaluation by an endocrinologist for decreased hormone levels and treatment as needed; monitor for adult-onset medical issues such as diabetes, high blood pressure, tremors, breast cancer, osteoporosis (thinning and weakening of the bones), and autoimmune conditions
Distal 16p11.2 deletion syndrome	Developmental delays or intellectual disability, obesity, macrocephaly (large head size)	Neurology evaluation to monitor for seizures, surveillance for neuropsychiatric concerns such as ASD and hyperactivity, encourage healthy eating habits and an active lifestyle
Distal 16p11.2 duplication syndrome	Poor weight gain, microcephaly, ASD	Feeding therapy, monitor for seizures and neuropsychiatric concerns such as ASD and ADHD
4p- syndrome (also called Wolf-Hirschhorn syndrome)	Poor growth before and after delivery, feeding difficulties, low muscle tone, various birth defects, seizures	Use of medications for seizures, feeding therapies, other care as needed for child's specific health concerns
Smith-Magenis syndrome	Developmental delays, ASD, behavioral problems (outbursts, impulsivity, aggression, self-injurious behaviors)	Consideration of medications for poor attention and/or hyperactivity, respite care and psychosocial support recommended for family members, multidisciplinary evaluations as needed
15q13.3 microdeletion syndromere	Developmental delays (mainly with speech), mild intellectual disability, behavioral problems, ASD	Evaluation of and treatment for heart defects, seizures, ASD, and schizophrenia
Duplication 15q syndrome (also called inversion duplication 15)	Low muscle tone, motor delays, intellectual disability, ASD, and epilepsy including infantile spasms	Consideration of medications to treat behavioral issues, monitoring for seizures and/or change in seizure type
22q13.3 deletion syndrome (also called Phelan McDermid syndrome)	Low muscle tone, absent to severely delayed speech, developmental delays and intellectual disability, ASD or autism-like behaviors	Use of assistive technology for communication; monitoring for seizures, lymphedema (swelling in the arms or legs), and dental damage (from chewing non-food items); avoid high temperatures or long periods in the sun due to decreased sweating, avoid sharp objects and tight clothes or shoes due to decreased awareness of pain

Angelman syndrome	Developmental delay, severe speech impairment, unsteady manner of walking, inappropriate happy demeanor (apparent after 12 months), microcephaly, seizures (usually starting before age 3)	Use of assistive technology for communication, specific medications for seizures, sedatives for nighttime wakefulness, surveillance and treatment of scoliosis as needed
17q12 deletion syndrome	Multicystic kidneys or other kidney conditions, non-insulin dependent diabetes starting in adolescence, and developmental delays or learning disability	Treatment and management of kidney anomalies and early onset of non-insulin dependent diabetes
17q12 duplication syndrome	Developmental delay, speech delay, intellectual disability or learning problems, autism, seizures (seen in 75%), eye or vision problems (seen in 33%)	Psychological evaluation and treatment as needed for ASD, schizophrenia, and behavioral abnormalities such as aggression and self-injury; evaluation for heart and kidney problems (rare cases)
3q29 deletion syndrome	Developmental delay and/or intellectual disability, speech delay, ASD	Evaluation, monitoring, and treatment of neuropsychiatric disorders; feeding therapy and/or consideration of a feeding tube; evaluation and management of heart defects, recurrent ear infections, and dental problems
5p- syndrome (also called cri-du-chat syndrome)	Unique high-pitched cat-like cry in infancy, developmental delay, low muscle tone, intellectual disability, microcephaly, poor growth	Treatment and management of hearing loss, vision concerns, and poor growth; specific medical care/treatment as needed based on the child's concerns
17q21.31 deletion syndrome (also called Koolen-de Vries syndrome)	Developmental delay, intellectual disability, seizures, various birth defects, low muscle tone, feeding difficulties	Feeding therapy; evaluation and treatment of hearing loss, vision problems, epilepsy, scoliosis, hip dislocation, and birth defects affecting the heart, kidneys, urinary tract system, or male reproductive organs
45,X (also called Turner syndrome)	Heart or kidney abnormalities, wide neck, low hairline at the back of the head, swelling of the hands or feet	Treatment and management of hearing loss, short stature, reproductive issues in adulthood, high blood pressure, and discussion of hormone replacement therapy; heart and kidney issues addressed as needed; monitoring for learning disabilities, social impairments, or ASD
17p11.2 duplication syndrome (also called Potocki-Lupski syndrome)	Low muscle tone, failure to thrive, developmental delay, heart defects, low blood sugar due to growth hormone deficiency	Multidisciplinary team care; surveillance and feeding therapy as needed for poor growth; monitor for behavioral concerns such as poor attention, hyperactivity, withdrawal, and anxiety; treatment of heart defects and other specific medical care/treatment as needed
2q37 deletion syndrome	Developmental delay, ASD, short fingers/toes, short stature, failure to thrive, low muscle tone	Multidisciplinary team care, surveillance for behavioral and cognitive changes, screening for kidney cysts at age 4 years and at puberty, screening for Wilms tumors as needed
1p36 deletion syndrome	Low muscle tone, developmental delays, and poor or absent speech, seizures, structural brain anomalies	Neurology evaluation, treatment, and monitoring for seizures and brain anomalies; feeding therapy and/or feeding tube; evaluation and treatment for possible heart issues, eye or vision problems, hearing loss, skeletal changes, and kidney or genitalia abnormalities
15q11.2 BP1-BP2 deletion	Developmental and language delay, general behavioral problems, palate abnormalities	Routine treatment of palate problems; monitor and treatment of less common features that include seizures, ASD, ADD, ADHD, and schizophrenia/paranoid psychosis
1q21 distal deletion syndrome	Developmental delays, mild-to-moderate intellectual disability, microcephaly, heart defects, eye abnormalities, skeletal changes, and other malformations	Evaluation and treatment of heart, eye, and skeletal problems; surveillance and intervention of psychiatric and behavioral problems, monitor for seizures (seen in 15%)
1q21 distal duplication syndrome	Developmental delays, mild-to-moderate intellectual disability, ASD, macrocephaly	Monitor for neuropsychiatric conditions such as ADHD and ASD
16p13.11 deletion syndrome	Developmental delay, intellectual disability, ASD, ADHD, and OCD	Monitor and interventions as needed for behavioral concerns and mental health changes, specific medical care/treatment as needed
9p deletion syndrome	Developmental delay, mild-to-moderate intellectual disability, abnormal development of the face and skull bones, hypotonia	Evaluation and treatment for possible heart defects, hernias, scoliosis, respiratory issues, frequent ear infections, or other specific medical care/treatment as needed
47,XXX	Developmental delays, ASD, ADHD	Surveillance for neuropsychiatric problems such as ADHD and ASD, evaluation and treatment for dental problems such as overcrowding and poor tooth enamel

Glossary:

ASD: autism spectrum disorder

ADHD: attention deficit-hyperactivity disorder

ADD: attention deficit disorder

Microcephaly: head size is smaller than expected

Macrocephaly: head size is larger than expected

Palate: roof of the mouth

Neuropsychiatric conditions: conditions that impact brain function, emotion, and mood.

These can include seizures, attention deficits, cognitive deficits, palsies, uncontrolled anger, migraine headaches, addictions, eating disorders, depression, anxiety, and others.