

# Vistara non-invasive prenatal screen



## Vistara identifies risk for conditions that may have otherwise gone undetected until after birth or into childhood

All conditions are inherited in an autosomal or X-linked dominant fashion, which means that if the mutation is present, the child will be affected by the condition and experience related symptoms.

Condition <sup>1</sup> <i>Gene(s)</i>	Clinical synopsis <sup>2,3</sup>	Cases caused by de novo mutations <sup>2,3</sup>	Ultrasound findings <sup>2,3</sup>			Clinical actionability	Detection rate for gene <sup>1</sup>
			None	Third trimester	Non-specific		
<b>Achondroplasia</b> <i>FGFR3</i>	The most common form of skeletal dysplasia; may cause hydrocephalus, delayed motor milestones, and spinal stenosis	80%		●	●	Labor and delivery management, monitor for spinal stenosis, early sleep studies to reduce risk of SIDS	>96%
<b>Alagille syndrome</b> <i>JAG1</i>	Affects multiple organ systems and may cause growth problems, congenital heart defects, and vertebral differences	50% to 70%	○		○	Symptom-based treatment	>86%
<b>Antley Bixler syndrome</b> <i>FGFR2</i>	A type of craniosynostosis; also causes premature fusion of the arm bones, blockage of the nasal passage, and permanently flexed or extended joints	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus	>96%
<b>Apert syndrome</b> <i>FGFR2</i>	A type of craniosynostosis; also causes abnormal formation of the fingers, toes, and vertebrae, and other organ anomalies	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus	>96%
<b>Cardiofaciocutaneous syndrome 1,3,4</b> <i>BRAF, MAP2K1, MAP2K2</i>	Causes abnormalities of the heart, face, skin, and hair; may cause developmental delays and intellectual disability	majority		●	●	Fetal echocardiogram	>96%
<b>CATSHL syndrome</b> <i>FGFR3</i>	Acronym stands for camptodactyly, tall stature, scoliosis, and hearing loss; may increase risk for intellectual disability	unknown	●			Early adoption of sign language and behavioral intervention	>96%
<b>CHARGE syndrome</b> <i>CHD7</i>	Acronym stands for coloboma, heart defects, atresia of the choanae, retardation of growth and development, genital abnormality, ear abnormalities; may cause hearing loss, developmental delays, and cleft lip and/or palate	majority	○	○	○	Early referral to endocrinology, adoption of sign language, and behavioral intervention	>91%
<b>Cornelia de Lange syndrome 1,2,3,4,5</b> <i>NIPBL, SMC1A, SMC3, RAD21, HDAC8</i>	Causes a range of physical, cognitive, and medical challenges	99%	○		○	Monitor for cardiac, GI, and limb comorbidities	>43% to >96%
<b>Costello syndrome</b> <i>HRAS</i>	Causes heart defects, intellectual disability, developmental delays, growth delays, and increased risk of malignant tumors	majority	○		○	Nasogastric or gastrostomy feeding, behavioral and medical intervention	>92%
<b>Crouzon syndrome</b> <i>FGFR2, FGFR3</i>	A type of craniosynostosis; also causes hearing loss and dental problems in some cases	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus, early adoption of sign language	>96%
<b>Ehlers-Danlos syndrome, classic, type VIIA, cardiac valvular form, type VIIB</b> <i>COL1A1, COL1A2</i>	Causes defects in connective tissue that can vary from mildly loose joints to life-threatening complications, such as aortic dissection	50%	●			Orthotic treatment, monitoring for vascular complications	>92%
<b>Epileptic encephalopathy, early infantile, 2</b> <i>CDKL5</i>	Causes seizures with secondary developmental delay	majority	●			Monitor and treat seizures	>84%

○ = some types or cases

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			None	Third trimester	Non-specific		
<b>Hypochondroplasia</b> <i>FGFR3</i>	Causes a mild form of dwarfism; may cause seizures with secondary developmental delay	up to 80%	●			Monitor and treat seizures	>96%
<b>Intellectual disability</b> <i>SYNGAP1</i>	Causes intellectual disability and developmental delays	~100%	●			Early behavioral interventions	>86%
<b>Jackson Weiss syndrome</b> <i>FGFR2</i>	A type of craniosynostosis; also causes foot abnormalities	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus	>96%
<b>Juvenile myelomonocytic leukemia (JMML)</b> <i>PTPN11</i>	A rare pediatric blood cancer; five-year survival is approximately 50%	unknown	●			Monitor bloodwork and medical intervention	>96%
<b>LEOPARD syndrome 1,2 (Noonan syndrome with multiple lentiginos)</b> <i>PTPN11, RAF1</i>	Similar to Noonan syndrome, with notable brown skin spots (lentiginos); causes short stature, heart defects, bleeding problems, and, in some cases, mild intellectual disabilities	unknown	◐		◐	Fetal echocardiogram	>96%
<b>Muenke syndrome</b> <i>FGFR3</i>	A type of craniosynostosis; may cause hearing loss, developmental delays, and cleft lip and/or palate	unknown		●		Fetal MRI, corrective surgery, early adoption of sign language, and behavioral intervention	>96%
<b>Noonan syndrome 1,3,4,5,6,8,9</b> <i>PTPN11, SOS1, RAF1, RIT1, KRAS, NRAS, SOS2, SHOC2, BRAF, MAP2K1, HRAS, CBL</i>	Causes short stature, heart defects, bleeding problems, and, in some cases, mild intellectual disabilities	25% to 70%	◐	◐	◐	Fetal echocardiogram, labor and delivery management, early assessment for learning differences	>92% to >96%
<b>Osteogenesis imperfecta, type I,II,III,IV</b> <i>COL1A1, COL1A2</i>	Causes fragile bones that break easily, often without an identifiable cause	more severe forms	◐	◐		Labor and delivery management, neonatal care, early recognition and treatment of fractures	>92%
<b>Pfeiffer syndrome type 1,2,3</b> <i>FGFR2</i>	A type of craniosynostosis; also causes hearing loss, intellectual disability, hand abnormalities, and may result in early death	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus, early adoption of sign language, and behavioral intervention	>96%
<b>Rett syndrome</b> <i>MECP2</i>	Causes a rapid regression in language and motor skills at 6 to 18 months of age; autism, seizures, and long QT syndrome are often present	>99%	●			Evaluate for cardiac risk, monitor and treat seizures, early medical and behavioral interventions	>78%
<b>Sotos syndrome 1</b> <i>NSD1</i>	Overgrowth syndrome; also causes developmental delays, intellectual disability, and behavioral problems	>95%	◐	◐		Fetal echocardiogram, fetal renal ultrasound, and early behavioral intervention	>47%
<b>Thanatophoric dysplasia, types I,II</b> <i>FGFR3</i>	A severe skeletal disorder that typically results in stillbirth or neonatal death due to respiratory failure	majority			●	Labor and delivery management	>96%
<b>Tuberous sclerosis 1,2</b> <i>TSC1, TSC2</i>	Causes benign tumor growth in many organ systems in the body that can be life-threatening; may also cause seizures and secondary developmental delays	66%	◐	◐		Fetal echocardiogram, postnatal MRI, medical and behavioral interventions	91%

◐ = some types or cases

#### References

1. Validation data. Baylor. 2016.
2. GeneReviews. <https://www.ncbi.nlm.nih.gov/books/NBK1116/>
3. Genetics Home Reference. <https://ghr.nlm.nih.gov/>

The tests described have been developed and their performance characteristics determined by the CLIA-certified laboratory performing the tests. These tests have not been cleared or approved by the U.S. Food and Drug Administration (FDA). Although FDA does not currently clear or approve laboratory-developed tests in the U.S., certification of the laboratory is required under CLIA to ensure the quality and validity of the tests.