Vistara non-invasive prenatal screen

natera™ | Vistara Single-gene NIPT

Vistara identifies probability 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[1] *Gene(s)*	Clinical synopsis[2,3]	Cases caused by de novo mutations[2,3]	Ultrasound findings[2,3]			Clinical actionability	Detection rate for gene[1]
			None	Third trimester	Non-specific		
Achondroplasia *FGFR3*	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%
Alagille syndrome *JAG1*	Affects multiple organ systems and may cause growth problems, congenital heart defects, and vertebral differences	50% to 70%	◐		◐	Symptom-based treatment	>86%
Antley Bixler syndrome *FGFR2*	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%
Apert syndrome *FGFR2*	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%
Cardiofaciocu-taneous syndrome 1,3,4 *BRAF, MAP2K1, MAP2K2*	Causes abnormalities of the heart, face, skin, and hair; may cause developmental delays and intellectual disability	majority		●	●	Fetal echocardiogram	>96%
CATSHL syndrome *FGFR3*	Acronym stands for camptodactyly, tall stature, scoliosis, and hearing loss; may increase risk for intellectual disability	unknown	●			Early adoptionof sign language and behavioral intervention	>96%
CHARGE syndrome *CHD7*	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%
Cornelia de Lange syndrome 1,2,3,4,5 *NIPBL, SMC1A, SMC3, RAD21, HDAC8*	Causes a range of physical, cognitive, and medical challenges	99%	◐		◐	Monitor for cardiac, GI, and limb comorbidities	53% to >96%
Costello syndrome *HRAS*	Causes heart defects, intellectual disability, developmental delays, growth delays, and increased risk of malignant tumors	majority	◐		◐	Nasogastric or gastronomy feeding, behavioral and medical intervention	>92%
Crouzon syndrome *FGFR2, FGFR3*	A type of craniosynostosis; also causes hearing loss and dental problems in some cases	more severe forms		●		Fetal MRI, avoid instru-mented delivery, corrective surgery, monitor for hydrocephalus, early adoption of sign language	>96%
Ehlers-Danlos syndrome, classic, type VIIA, cardiac valvular form, type VIIB *COL1A1, COL1A2*	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%
Epileptic encephalopathy, early infantile, 2 *CDKL5*	Causes seizures with secondary developmental delay	majority	●			Monitor and treat seizures	>84%

◐ = some types or cases

Condition[1] *Gene(s)*	Clinical synopsis[2,3]	Cases caused by de novo mutations[2,3]	Ultrasound findings[2,3]			Clinical actionability	Detection rate for gene[1]
			None	Third trimester	Non-specific		
Hypochondroplasia *FGFR3*	Causes a mild form of dwarfism; may cause seizures with secondary developmental delay	up to 80%	●			Monitor and treat seizures	>96%
Intellectual disability *SYNGAP1*	Causes intellectual disability and developmental delays	~100%	●			Early behavioral interventions	>86%
Jackson Weiss syndrome *FGFR2*	A type of craniosynostosis; also causes foot abnormalities	more severe forms		●		Fetal MRI, avoid instrumented delivery, corrective surgery, monitor for hydrocephalus	>96%
Juvenile myelomonocytic leukemia (JMML) *PTPN11*	A rare pediatric blood cancer; five-year survival is approximately 50%	unknown	●			Monitor bloodwork and medical intervention	>96%
LEOPARD syndrome 1,2 (Noonan syndrome with multiple lentigines) *PTPN11, RAF1*	Similar to Noonan syndrome, with notable brown skin spots (lentigines); causes short stature, heart defects, bleeding problems, and, in some cases, mild intellectual disabilities	unknown	◐		◐	Fetal echocardiogram	>96%
Muenke syndrome *FGFR3*	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%
Noonan syndrome 1,3,4,5,6,8,9 *PTPN11, SOS1, RAF1, RIT1, KRAS, NRAS, SOS2, SHOC2, BRAF, MAP2K1, HRAS, CBL*	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%
Osteogenesis imperfecta, type I,II,III,IV *COL1A1, COL1A2*	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%
Pfeiffer syndrome type 1,2,3 *FGFR2*	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%
Rett syndrome *MECP2*	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%
Sotos syndrome 1 *NSD1*	Overgrowth syndrome; also causes developmental delays, intellectual disability, and behavioral problems	>95%	◐	◐		Fetal echocardiogram, fetal renal ultrasound, and early behavioral intervention	>47%
Thanatophoric dysplasia, types I,II *FGFR3*	A severe skeletal disorder that typically results in stillbirth or neonatal death due to respiratory failure	majority			●	Labor and delivery management	>96%
Tuberous sclerosis 1,2 *TSC1, TSC2*	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% to >96%

◐ = some types or cases

References
1. Validation data, Baylor. 2020.
2. GeneReviews. https://www.ncbi.nlm.nih.gov/books/NBK1116/
3. Genetics Home Reference. https://ghr.nlm.nih.gov/