GeneSafe™ DE NOVO

identifies fetal conditions that could be **missed by traditional prenatal screening.**

GENE	SYNDROMIC DISORDERS
JAG1	Alagille syndrome
CHD7	CHARGE syndrome
HDAC8	Cornelia de Lange syndrome 5
NIPBL	Cornelia de Lange syndrome 1
MECP2	Rett syndrome
NSD1	Sotos syndrome 1
ASXL1	Bohring-Opitz syndrome
SETBP1	Schinzel-Giedion syndrome
SIX3	Holoprosencephaly
	SINDROME DI NOONAN
BRAF	Cardiofaciocutaneous syndrome 1
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL)
KRAS	Noonan syndrome/cancers
MAP2K1	Cardiofaciocutaneous syndrome 3
MAP2K2	Cardiofaciocutaneous syndrome 4
NRAS	Noonan syndrome 6/cancers
PTPN11	Noonan syndrome 1/ LEOPARD syndrome/cancers
PTPN11	Juvenile myelomonocytic leukemia (JMML)
RAF1	Noonan syndrome 5/LEOPARD syndrome 2
RIT1	Noonan syndrome 8
SHOC2	Noonan syndrome-like disorder with loose anagen hair
SOS1	Noonan syndrome 4

GENE	SKELETAL DISORDERS
COL2A1	Achondrogenesis, type II or hypochondrogenesis
FGFR3	Achondroplasia
	CATSHL syndrome
	Crouzon syndrome with acanthosis nigricans
	Hypochondroplasia
	Muenke syndrome
	Thanatophoric dysplasia, type I
	Thanatophoric dysplasia, type II
COL1A1	Ehlers-Danlos syndrome, classic
	Ehlers-Danlos syndrome, type VIIA
	Osteogenesis imperfecta, type I
	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form
	Ehlers-Danlos syndrome, type VIIB
	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
	CRANIOSYNOSTOSIS SYNDROMES
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
	Apert syndrome
	Crouzon syndrome
	Jackson-Weiss syndrome
	Pfeiffer syndrome type 1
	Pfeiffer syndrome type 2
	Pfeiffer syndrome type 3

GeneSafe™ detects de novo mutations in 25 genes causing 44 different genetic disorders. The genetic conditions screened by this innovative test often occur in the absence of a family history of the condition. This is a paradigm shift in prenatal screening. GeneSafe™ screens for de novo mutations that cannot be detected by standard carrier screening, as these mutations are not present on the parents. The genetic disorders screened by GeneSafe™ can cause **skeletal dysplasias, cardiac defects,** [1-2-3] **multiple congenital anomalies,** [4-5] **autism,** [6] **epilepsy** [7] **and/or intellectual disability.** [8-9]

1. Homsy J, et al. *Science. 2015;350:1262–6.* **2.** Zaidi S, et al. *Nature. 2013;498:220–3.* **3.** Sifrim A, et al. *Nat Genet. 2016;48:1060–5.* **4.** Ng SB, et al. *Nat Genet. 2010;42:790–3.* **5.** Hoischen A, et al. *Nat Genet. 2011;43:729–31.* **6.** O'Roak BJ, et al. *Nat Genet. 2011;43:585–9.* **7.** Allen AS, et al. *Nature. 2013;501:217–21.* **8.** de Ligt J, et al. *N Engl J Med. 2012;367:1921–9.* **9.** Rauch A, et al. *Lancet. 2012;380:1674–82.*