Why MedGenome

- Unmatched experience of 350,000+ genomes/exomes
- South Asia largest database with 5,000,000+ variants
- Recommended by 4000+ hospitals & trusted by 10000+ clinicians
- Consistent proficiency testing through "External Quality Monitoring Programs"
- Two levels of analysis and review by molecular and clinical geneticists

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Talk to your doctor immediately incase of any family history.

MedGenome offers free Expert Genetic Counselling to understand the risk, test details & results.

MEDGENOME

Talk to the Experts

1800 103 3691

diagnostics@medgenome.com

www.medgenome.com

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COLLEGE of AMERICAN PATHOLOGISTS

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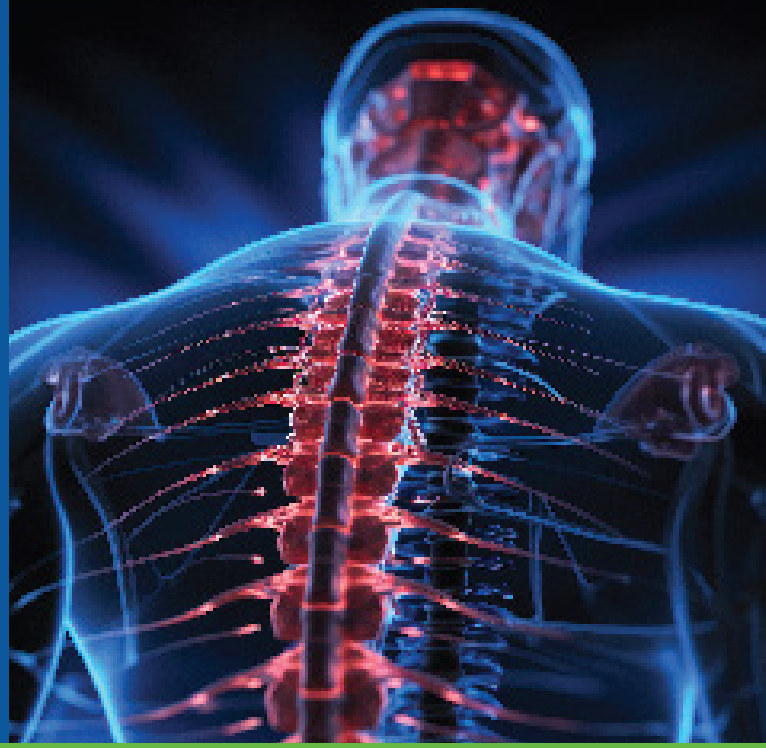
MEDGENOME | ACTIA Inherited Disease Genetics

Spinal Muscular Atrophy (SMA)

Inherited disorder affecting the motor neuron that control voluntary muscle movement



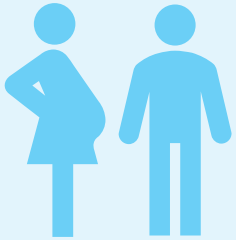
SMA affects

~1 in 10,000

live births worldwide

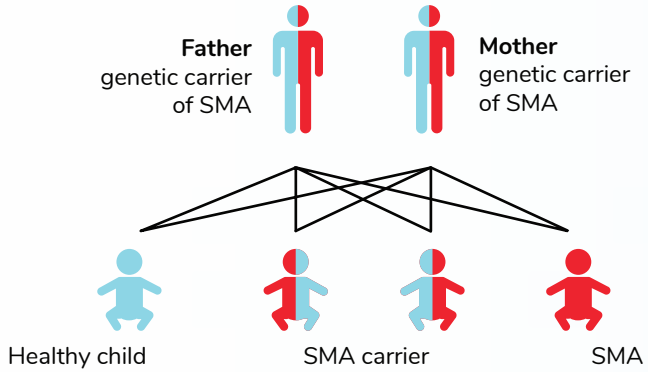
~1 in 54

People carry the genetic defect & can affect any race or sex



When both parents are carriers, baby has a **25% chance** of having SMA

Inheritance Pattern



SMN1 gene	
Proportion of Pathogenic Variants detectable	
Sequence Analysis	Gene-targeted deletion / duplication analysis
2%-5%	95%-98%
SMN2 evaluation for prognostics	

Type 1 (severe)

~60%, at birth or within an infant's first 6 months of life

Type 2 (intermediate)

between six months and 18 months old

Type 3 (mild)

appear after a child's first 18 months of life

Type 4 (adult)

The rare adult form in the mid-30s

Test Details

Test Code	Test Name	Methodology	TAT
MGM144	Spinal Muscular Atrophy (SMN1) gene analysis	Sanger Sequencing	21 Working days
MGM143	Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication analysis	MLPA	14 Working days

New therapeutic approaches have become available in the past few years

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