What is SMA?

SMA stands for Spinal Muscular Atrophy. Some people refer to SMA as ALS in babies. It weakens the muscles used to breathe, swallow, walk, sit and even smile.

- It is the # 1genetic killer of infants
- 1in 50 people are carriers of SMA
- 1in 8,000 people are affected
- Affected individuals have zero copies of
- the SMN1 gene
- Most people don't know their carrier status
- There are 4 types of SMA
- Type 1 is the most severe
- SMA is pan-ethnic

The mind remains unaffected with SMA. There is currently no treatment or cure for this degenerative neuromuscular disease.

Early detection is key. Know your carrier status. It's as easy as asking your doctor for a carrier screening test.

Many labs offer genetic screening. One option is through Counsyl.

Visit www.counsyl.com and look for the Family Prep Screen or www.liverhysstrong.org/act for additional options.

LIVERHYSSTRONG FOUNDATION
www.liverhysstrong.org
info@liverhysstrong.org
415 606.7424

SMA Types

SMA Types vary by milestones reached. Typically, stronger individuals have more copies of the backup gene, SMN2.

Type	Age of Onset	Symptoms
1 (Werdnig-Hoffman disease)	0-6 months	• Most severe form • Loss of neck control • Hypotonia (floppy limbs & frog legs) • Poor reflexes • Tongue tremors • Excessive drooling • Poor weight gain due to swallowing problems • Breathing from the diaphragm • Occurs in 70% of SMA cases
2	6-18 months	• Trouble breathing • Can sit without support • Unable to stand or walk unassisted
3 (Kugelberg-Welander disease)	Early childhood (24 months)-adolescence	• Abnormal gait • Difficulty running, climbing steps or rising from a chair • Fine tremor of the fingers
4	Adulthood	• Mild motor impairment • Mild breathing problems

Inheritance Pattern

SMA has an autosomal recessive inheritance pattern. Carriers do not show symptoms. It only takes one parent to pass the mutated gene to a carrier child.

When two carriers have a child, the probability of passing the disease is:

Affected: 1 in 4

Carrier: 1 in 2

Unaffected: 1 in 4

This is with each and every pregnancy.

SMN2 is a backup gene with limited function.