

SMA STATE FACT SHEET



California

Est. babies born with SMA annually: 45
Est. individuals living with SMA: 1,448
Est. number of SMA carriers: 724,904

Estimates for incidence, prevalence, and carriers are based on 2015 birth and population data for the state of California.

About Spinal Muscular Atrophy (SMA)

- A progressive neurodegenerative disease that robs people of physical strength by affecting the motor nerve cells in the spinal cord, impeding their ability to walk, swallow, and in the most severe cases the ability to breathe.
- Leading genetic cause of death for infants today, affecting approximately 1 in 11,000 babies; there are four types of SMA (more info on reverse).
- Disease is caused by a mutation in the survival motor neuron gene 1 (*SMN1*); without enough of the SMN protein nerve cells cannot function properly and eventually die, leading to debilitating and often fatal muscle weakness.
- Some estimates indicate that approximately 12,000 Americans are affected with one of the four types of SMA. About 1 in every 50 Americans is a genetic carrier, and SMA can affect any race or gender.

New Hope for Treating SMA

- December 23, 2016: FDA approves first-ever treatment for SMA – SPINRAZA™.
- Intrathecal injection (into the lower back, directly into the spinal canal).
- Four “loading” doses in the first two months, then a maintenance dose once every four months thereafter.
- Increases the production of the survival motor neuron (SMN) protein, which supports the nerve cells that control muscles.
- Preliminary results of the continuing NURTURE clinical trials for SPINRAZA™ demonstrated a clear benefit for the pre-symptomatic treatment of SMA patients. Infants who were diagnosed before the onset of symptoms who received SPINRAZA™ achieved unprecedented motor milestones of sitting, standing, and walking, never before seen for infants with SMA type I.

California Cure SMA Information

- More than 435 families in the Cure SMA community network in the state.
- Served by the Northern California and Southern California Cure SMA Chapters.



Type I

- Most severe, most common (approx. 60% cases).
- Often fatal; morbidity between 8 and 11 months of age.
- Diagnosed in first 6 months of life.
- Muscle weakness, trouble breathing, coughing, and swallowing; normally need breathing machines and feeding tubes.

Type II

- Approx. 30% of SMA cases.
- Usually diagnosed after 6 months of age following failure to achieve certain motor function milestones.
- Able to sit up without help; however, may need assistance sitting; unable to walk and require wheelchair for mobility.

Type III

- Approx. 10% of SMA cases.
- Diagnosis typically after 18 months of age; can be diagnosed into late teenage years.
- Initially may be able to walk, but can deteriorate over time; use of wheelchair common.

Type IV

- Very rare; less than 1% of SMA cases
- Typically diagnosed in adulthood (after age 35); some diagnoses as early as 18 years-old
- Mild motor impairment