

Disease

Spinal muscular atrophy-1¹

Gene

SMN1

Inheritance	AR
Current Treatments	risdiplam , nusinersen, gene therapy (onasemnogene abeparvovec-xioi)
Evidence for Treatment	guideline ^{2, 3}
Non-Molecular Testing Availability	yes
Non-Molecular Test	electromyography and nerve conduction studies ⁴
Disease Frequency (per 100,000)	10 ⁵
Genes Included in Disease Frequency	SMN1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Spinal+Muscular+Atrophy&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1352/>
- ² <https://pubmed.ncbi.nlm.nih.gov/29290580/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29305137/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/25346245/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5496354/>