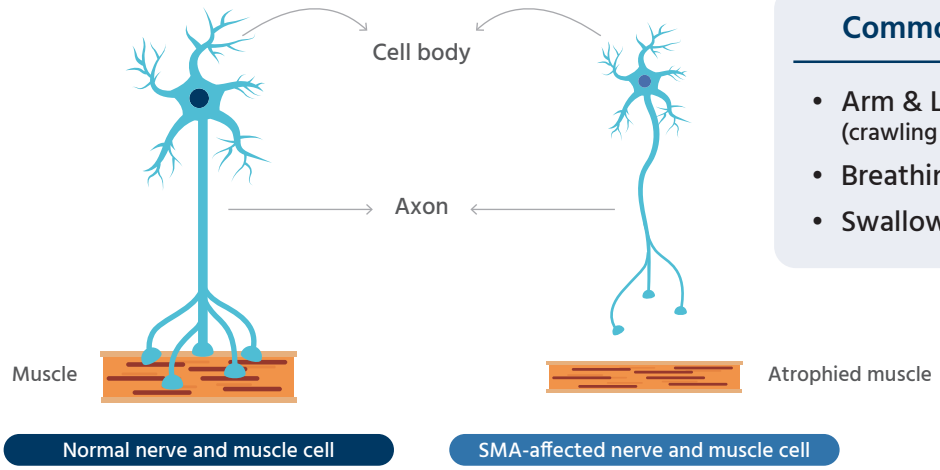
Spinal Muscular Atrophy
diagnosis & carrier screening test

SMN1, SMN2 Del/Dup

What is Spinal Muscular Atrophy?



Common symptoms

- Arm & Leg weakness
  (crawling & walking difficulties)
- Breathing difficulties
- Swallowing problems

SMA (Spinal Muscular Atrophy) is a hereditary neuromuscular disorder caused by mutations in the SMN gene, which is responsible for maintaining motor neurons.
Due to insufficient production of motor neurons, individuals with SMA experience genetic neuromuscular conditions.

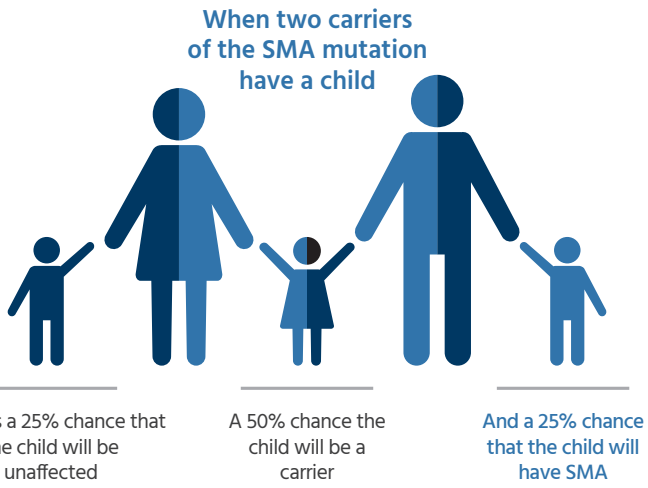
How can we test for SMA?

Among the SMN1 and SMN2 genes associated with this disease, the SMN1 gene is a major gene related to disease development. 95% of patients are homozygous for the SMN1 gene deletion mutation, and the remaining 5% are heterozygous for the deletion mutation and point mutation.

Through SMN1, SMN2 del/dup test, we can confirm deletion and duplication of exon 7 and 8 of SMN1&SMN2 genes using the MLPA (Multiplex Ligation dependent Probe Amplification) method.

Purposes of the test

1. SMA diagnosis when the patient has suspicious symptoms
2. SMA carrier screening for parents to see if they are at increased risk of having a child affected with SMA



Spinal Muscular Atrophy
diagnosis & carrier screening test

SMN1, SMN2 Del/Dup

Importance of SMN1, SMN2 del/dup test

Early diagnosis and prompt initiation of the treatment is a key factor affecting the treatment response.

- Clinical Suspicion
- Clinical Diagnosis

+ Genetic test

→ ✓ Confirmation of SMN1 deletion
✓ Confirmation of SMN2 copy number

✓ Various SMA treatments can be applicable based on the test results.

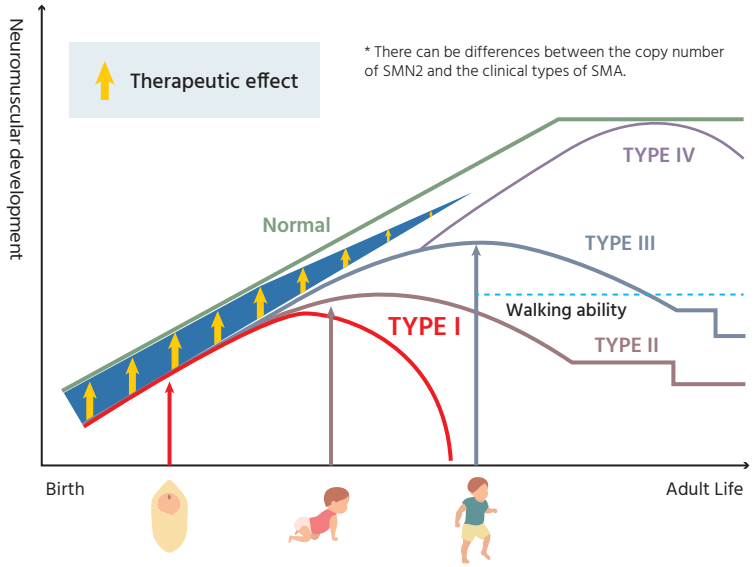


Fig1. European Journal of Human Genetics volume 27, pages1774–1782(2019)

Especially, **SMA Type 1** symptoms progress rapidly and often become severe

Early diagnosis
Prompt initiation of treatment

The greatest chance to achieve normal functioning

• Treatments

- SPINRAZA®
- Zolgensma®
- Evrysdi®

Service features

Test	SMN1, SMN2 del/dup	**Code**	OS094
Specimen	EDTA WB 3 ml	**TAT**	17 days
Method	MLPA (Multiplex Ligation-dependent Probe Amplification)	**Sample Storage**	Room temperature (Refrigerated is recommended.)
Test description	Spinal muscular atrophy(SMA) is one of the most common autosomal recessive diseases as a neuromuscular disease characterized by loss of symmetrical proximal muscle strength due to degeneration of anterior horn cells of the spinal cord or brain stem. Among the SMN1 and SMN2 genes associated with this disease, the SMN1 gene is a major gene related to disease development. 95% of patients are homozygous for the SMN1 gene deletion mutation, and the remaining 5% are heterozygous for the deletion mutation and point mutation. This test is a test to confirm deletion and duplication of exon 7 and 8 of SMN1&SMN2 genes using the MLPA (Multiplex Ligation dependent Probe Amplification) method.		
Caution & Limitation	• If there is a nucleotide sequence mutation at the MLPA probe attachment site, a false-positive result may be shown. • It is impossible to distinguish the normal individuals who have one SMN 1 gene in both alleles (1+1) normally and the carriers who have two SMN1 genes in one allele (2+0). There is no problem in diagnosing the patient, but there is a possibility of a false-negative result that reports carriers (2+0) as normal when tested to screen the carriers.		

GC Genome
Cell Center Bldg 1F, 107, Ihyeon-ro 30beon-gil, Giheung-gu, Yongin-si, Gyeonggi-do, Republic of Korea (16924)
Online : www.gc-genome.com| **Contact** : +82) 31-280-9910

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS