Spinal Muscular Atrophy Type 2

This information sheet briefly explains the cause, effects, and management of Spinal Muscular Atrophy (SMA) Type 2. It includes sources of further information and support. It is for the families of children diagnosed with SMA Type 2. It may also be useful for healthcare and other professionals.

The glossary at the end further explains the words that appear in bold font.

More information on SMA Type 2 and sources of support is available from SMA Support UK's route map for SMA Type 2: www.routemapforsma.org.uk

SMA Type 2 is a complex condition; there is a lot of information to take in, and every child with SMA is different. Your child's medical team will always be happy to go over any of this with you.

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a **rare**, **genetically inherited neuromuscular** condition. SMA may affect crawling and walking ability, arm, hand, head and neck movement, breathing and swallowing. SMA is often grouped into 'Types'. Types of SMA are based on the age at which symptoms first appear and what physical 'milestones' a baby or child is likely to achieve. Milestones can include the ability to sit, stand, or walk.

There are four main types of SMA: Types 1, 2 and 3 appear in childhood; Type 4 appears in adulthood and is also known as Adult Onset SMA.

These 'Types' are not rigid categories. There is a wide spectrum of severity both between the different types of SMA and between children, young people, and adults within each type.

There are also other, even rarer, forms of SMA with different **genetic** causes including SMA with Respiratory Distress (SMARD), Spinal and Bulbar Muscular Atrophy (SBMA), and Distal SMA (DSMA).

What causes SMA?

Usually, electrical signals from our brain are sent down our **spinal cord** along our **nerve cells** and through to our muscles. This makes it possible for us to consciously contract our muscles and to make them move.

SMA affects a particular set of **nerve cells** called the lower **motor neurones**[1] which run from the **spinal cord** out to our muscles. The lower motor neurones carry messages that make it possible for us to move the muscles we use to crawl and walk, to move our arms, hands, head, and neck, and to breathe and swallow.

For our lower **motor neurones** to be healthy, we need to produce an important **protein** called the **Survival Motor Neuron (SMN) protein**. Our ability to do this is controlled by a **gene** called **Survival Motor Neuron 1 (*SMN1*)**[2]**.**

We all have two copies of this ***SMN1* gene**, one from each parent. People who have two faulty copies of the *SMN1* gene have SMA. People who have one faulty copy of the *SMN1* gene are **carriers** of SMA. Carriers do not have SMA or any symptoms of SMA. People who have two healthy copies of the *SMN1* gene do not have SMA and are not carriers.

SMA is passed from parents to their children through their ***SMN1* genes**. When two people who are **carriers** have a child together, their child may inherit two faulty *SMN1* genes, one from each parent. If this happens, then their child will have SMA.

Having two faulty ***SMN1* genes** means that a child is only able to produce very low amounts of the **SMN protein**. This causes their lower **motor neurones** in their **spinal cord** to deteriorate. Messages from their spinal cord do not efficiently get through to their muscles, which makes movement difficult. Their muscles waste due to lack of use and this is known as muscular **atrophy**.

For more information on 'The Genetics of Spinal Muscular Atrophy' please see: http://www.smasupportuk.org.uk/the-genetics-of-sma

What is SMA Type 2?

SMA Type 2 is sometimes called intermediate SMA or chronic infantile SMA. The symptoms of muscle weakness and floppiness (low tone / **hypotonia**) usually appear between 7 and 18 months of age.

Each child with SMA Type 2 is different. Some children will sit independently whilst others will require some support. Usually children with SMA Type 2 will need supportive aids for standing and a wheelchair to get around.

Sometimes doctors try to indicate the degree of severity within SMA Type 2 by using a decimal classification[3], for example, 2.1, 2.2, 2.5, 2.9. If you have any queries regarding this please speak to your child's medical team.

Though this is a serious **inherited neuromuscular** condition that may shorten life expectancy, improvements in care standards mean that the majority of people can live long, fulfilling and productive lives.

How is SMA Type 2 diagnosed?

A doctor will **diagnose** SMA Type 2 after taking a medical history, physically examining your child, and by taking a blood sample for **DNA** testing. The blood sample is tested for a **deletion mutation** in the **survival motor neurone 1 (*SMN1*) gene** on **chromosome** 5. The result of this test is usually available within 2 – 4 weeks.

If there is any uncertainty about the diagnosis, further muscle tests such as an **electromyogram (EMG)** or a **muscle biopsy** may be discussed, but these are not usually needed to confirm SMA.

Is there a treatment or cure for SMA Type 2?

Although there is currently no cure for SMA, this does not mean that nothing can be done. Symptoms can be managed so that your child can achieve their maximum mobility, independence, and quality of life.

What are the effects of SMA Type 2?

This section describes the effects of SMA Type 2 in general terms. But, it's important to remember that each child with SMA Type 2 is affected differently and the severity of the condition varies from child to child.

Children's muscle weakness is usually the same on both sides of their body (**symmetrical**). The muscles closest to the centre of their body (**proximal** muscles) are usually more severely affected than the muscles furthest from the centre of their body (**distal** muscles). This can make it difficult for children with SMA Type 2 to lift their arms and legs but they will still be able to use their hands and fingers. Generally, children with SMA Type 2 find that their legs are weaker than their arms.

As your child grows it may be difficult for their muscles to keep up with their daily activities. If your child has been able to, for example, crawl or roll, they may lose this ability as they get older. They may also become weaker after infections and at times of major growth spurts such as puberty. SMA doesn't affect a child's sexual or intellectual[4] development.

Because a child with SMA Type 2 has weak **respiratory** muscles it can be difficult for them to cough effectively. This can make them more vulnerable to respiratory (chest) infections.

In SMA Type 2 the muscles supporting the **spinal** column are weakened. This means that most children will develop a sideways curvature of their spine[5] (**scoliosis**). Also, because the condition reduces children's ability to move, some joints may become tight (**contracture**) restricting their range of movement.

SMA Type 2 can weaken children's chewing and swallowing muscles (**bulbar muscles**). Some children find that their tongue or shoulder muscles twitch (**fasciculation**) and they may have a slight tremor in their hands[6]. SMA Type 2 doesn't affect bladder and bowel control but

children will need help, for example, with transferring from their wheelchair to the toilet, dressing, and undressing.

What healthcare and support is needed for SMA Type 2?

Your child should receive care from a multidisciplinary healthcare team, which can feel like an overwhelming number of people but they all have an important role to play. You will have contact with specialists in **neuromuscular** conditions, **respiratory** medicine, **orthopaedics**, physiotherapy, occupational therapy, speech and language therapy, dietetics, and a hospital or community consultant paediatrician. If possible one of the team should be your keyworker whose job it is to help co-ordinate services for your family. You can find out more about how these people help in our information sheet 'Who's Who of Professionals': http://www.smasupportuk.org.uk/whos-who-of-professionals

Children with SMA Type 2 should be seen by their medical team regularly to measure any change in their health and to offer advice and interventions at the right time. The aim is to enable your child to remain healthy and enjoy a good quality of life.

At every appointment with your child's medical team you should be given time to ask questions and then jointly decide what support is best for your child.

- Breathing

Children with SMA Type 2 can have breathing problems such as **hypoventilation** and their muscle weakness can lead to an increased risk of **respiratory** (chest) infections. Some may need help with their breathing and coughing. Your child's medical team will advise you on what is best for your child.

You can read an overview of what good **respiratory** management involves in the booklet 'Standards of Care for Spinal Muscular Atrophy – the family guide' [7]: http://www.treat-nmd.eu/care/sma/family-guide/ As each child is affected differently it's important to discuss any queries you have with your child's medical team.

- Eating, drinking and nutrition

Children with SMA Type 2 sometimes have difficulty with their chewing, swallowing, and nutrition.

Your child's health visitor, consultant, speech and language therapist, or dietitian will be able to provide advice and support on eating and nutrition. Occupational therapists and physiotherapists may also advise on positioning, seating, or arm supports to help your child to eat independently.

Your child may have problems putting on weight if eating becomes tiring for them or if they have illnesses or infections. Equally, your child may become overweight due to their reduced mobility. If this happens the extra weight can increase the stress on muscles, bones and joints,

making physical activity even more difficult. A dietitian will be able to advise on a healthy diet that will suit your child.

If your child has difficulty swallowing there is a risk that they may inhale liquids or food into their lungs (**aspirate**) which can cause chest infections. If this is happening, or your child is not putting on weight, your child's medical team may suggest alternative ways for them to take in food. This may involve your child's food going through a tube directly into their stomach (**gastrostomy (G) tube**).

Children with SMA Type 2 can become constipated which may cause discomfort and can make breathing more difficult. You can discuss how to manage this with your child's medical team.

Muscle weakness may make it difficult for your child to open their mouth widely. This can cause problems with eating, teeth cleaning, and dental care. Regular dental check-ups and getting help with these sorts of problems early may help to prevent complications such as **aspiration**[8], when food gets into the lungs.

- Posture, movement and mobility

SMA Type 2 will mean that your child will have difficulties with their posture, movement, and mobility. They will need their own exercise routines designed by their physiotherapist to help with this. Routines may include exercises to help maintain their range of motion, reduce any discomfort, stretch any tight muscles, and prevent **contractures**. Regular gentle stretching of their tight joints can help to reduce the pain that contractures can cause. If your child does have any pain, do talk to their doctor and physiotherapist. Your child might enjoy doing these exercises in the bath, or a swimming or hydrotherapy pool as the warm water aids buoyancy.

Regular moderate exercise will also help your child to maintain their fitness and stamina and activities such as swimming and horse riding can be adapted to match their physical ability.

Although your child will lose muscle strength over time, it is important that they maintain activities like supported standing for as long as possible. Standing is good for breathing, blood circulation, bladder, bowels, bones and joints.

As your child's physical abilities change, an occupational therapist will advise what sort of seating will give them the best, most comfortable support. This will make it easier for them to play with toys, eat independently, and join in at home and at school.

A physiotherapist will assess your child and provide appropriate equipment to support their standing and positioning. Some children benefit from having splints (sometimes called **orthoses**) for support. Types of orthoses include ankle foot orthoses (AFOs) and knee, ankle, foot orthoses (KAFOs). These will be made specifically for your child by an orthotist who will explain how they will help.

Your child's physiotherapist and occupational therapist will be able to advise you about powered wheelchairs. This should be at around the time that your child would have been learning to walk. This equipment will mean that they will be able to explore and join in much

more easily and this will help with their physical, emotional, social, and educational development[9].

As already mentioned, most children with SMA Type 2 develop a sideways curvature of their spine (**scoliosis**). It is important that the medical team monitors your child regularly so that any increase in curvature is noticed early. The degree of the curvature and your child's age will be factors in deciding how to manage this. Initially this may be with a **spinal** brace or jacket but surgery to correct scoliosis may be recommended if the scoliosis is contributing to breathing difficulties, is preventing comfortable sitting, or if the curvature has progressed beyond a certain point.

You may be provided with a sleep system which will support your child's back, arms, and legs, and make sleeping more comfortable. Your occupational therapist can advise you about sleep systems.

Occupational therapists can also give you advice about other adaptations and equipment that will help with your child's everyday activities such as writing, playing, washing, dressing, cooking and eating, both at home and at school. With appropriate encouragement, adaptations, and support, your child will be able to lead as fulfilling a life as their friends.

- Emergency health plans

Your child's medical team may work with you to develop an emergency health plan. This records the treatment you wish your child to receive if there is an emergency or if their health deteriorates. You should have your own copy so that you can give it to hospital services if you are away from your home area. The plan can be reviewed and you can change your mind about its contents at any time.

What other help is available?

A diagnosis of SMA Type 2 with all its complexity has an enormous impact on families. It's important for you and your child to have emotional support and plenty of time to talk and ask questions. This can be with members of your child's medical team, your local G.P., health visitor, social worker, psychologist or a counsellor.

To enable your child to fully participate in activities at home, school, and in their community, you will need information, advice and support on mobility, education, equipment and sources of funding that will aid their inclusion. You can find out more by talking to your child's healthcare team, Spinal Muscular Atrophy Support UK (SMA Support UK), and the other people and agencies listed in this leaflet and on the SMA Support UK route map for SMA Type 2: www.routemapforsma.org.uk

SMA Support UK can provide information and support to families affected by SMA in the UK. Our Outreach Workers are able to visit you at home and can discuss with you the health, social, educational, financial, and care support that you and your child may be entitled to. We can also put you in touch with our Peer Support Volunteers who have personal experience of

living with SMA Type 2. Information about these services is available on our website: www.smasupportuk.org.uk or please phone us on 01789 267 520 or email: supportservices@smasupportuk.org.uk

Muscular Dystrophy UK also provides information, support, and advocacy services, including grants towards specialist equipment, for people affected by a range of **neuromuscular** conditions. Their website is: www.musculardystrophyuk.org or you can phone them on 0800 652 6352 or e-mail: info@musculardystrophyuk.org

Regional care advisors, and sometimes **neuromuscular** nurse specialists, are attached to NHS neuromuscular clinics in various regions of the UK. They provide support and information to children and adults with muscle diseases and their families. They link up with other professionals and services so that people receive the local health and social support they need. Regional care advisors' contact details are available on Muscular Dystrophy UK's website: http://www.musculardystrophyuk.org/get-the-right-care-and-support/people-and-places-to-help-you/care-advisors/

Children's hospices throughout the UK also offer a wide range of services and support to children and families. Some also offer short breaks. Details of hospice services are available from 'Together for Short Lives' on 0808 8088 100 and more information is available on their website: www.togetherforshortlives.org.uk

- Financial Support

Families living in the UK may be eligible for a number of financial benefits to help towards the cost of providing the extra care their child may need. This does depend on your individual circumstances.

For further information about financial benefits visit the Gov.UK website www.gov.uk and look at the sections 'Benefits' and 'Carers and Disability Benefits'. The Department of Work and Pensions (DWP) can be contacted on: 0345 608 8545.

Contact a Family provide information and support to families who have a child with a disability. This includes information on benefits and grants. They can be contacted on 0808 808 3555 or through their website: www.cafamily.org.uk

Disability Rights UK publishes free factsheets on a range of benefits and the 'Disability Rights Handbook' annually. They do not have a helpline. For further information visit: www.disabilityrightsuk.org

Turn2Us is a charity which helps people access money available to them through welfare benefits, grants, and other help. They can be contacted on 0808 802 2000 or through their website: www.turn2us.org.uk

Your health visitor, **neuromuscular** care advisor, family support worker, social worker or outreach worker may be able to help you with applications for financial benefits.

There are also a number of charities that may assist you with the cost of general household goods, specialist equipment, and holidays / days out. Please contact SMA Support UK for more information or see the SMA Type 2 route map: www.routemapforsma.org.uk

- Genetic Counselling

As a parent with a child with SMA you should be offered a referral for **genetic counselling** to help you understand how SMA is passed on and what the chances are of other people in your family being affected. Genetic counselling also provides you with the opportunity to discuss your choices for any future pregnancies.

As your child and any siblings grow up, they can also ask for genetic counselling, particularly if they are considering having children.

For more information on the genetics of SMA, the chances of having a child with SMA and the tests that can be carried out, please see our leaflet 'The Genetics of Spinal Muscular Atrophy': http://www.smasupportuk.org.uk/the-genetics-of-sma

For more information on 'Future Options in Pregnancy' please see: http://www.smasupportuk.org.uk/future-options-in-pregnancy

What does the future hold?

To find out more about SMA research go to the research pages at: www.smasupportuk.org.uk/research

As new treatments for SMA are being developed they need to be tested in **clinical trials** but because SMA is a **rare** condition it can sometimes take years to find enough patients for a clinical trial. The UK SMA Patient Registry is a database of **genetic** and **clinical** information about people affected by SMA which aims to speed up this process. The Registry also helps specialists gain more knowledge about the condition and the number of people affected by SMA. This information helps to develop and improve worldwide standards of care for people with SMA. You can find out more by looking at their website: www.treat-nmd.org.uk/registry e-mailing: registry@treat-nmd.org.uk or phoning: 0191 241 8617.

Further Resources

SMA Support UK Information

Hard copies of the following leaflets can be requested from SMA Support UK on 01789 267 520 or downloaded from the website: www.smasupportuk.org.uk

- Spinal Muscular Atrophy – Information for Families
- Who's Who of Professionals
- Genetics of Spinal Muscular Atrophy
- Future Options in Pregnancy
- Information and Support
- The Outreach Service

Online resources

- SMA Type 2 Route Map: www.routemapforsma.org.uk

Other publications

- Smasheroo – an illustrated book for young children affected by SMA Type 2 or SMA Type 3
- Tilly Smiles – Tilly has SMA Type 2 and she and her family have written this book to inspire others

Standards of Care for Spinal Muscular Atrophy (TREAT-NMD)

This booklet describes best practice management and treatment for the more common forms of SMA. It is used by doctors but is also available to families. A hard copy can be requested from SMA Support UK. It can also be downloaded from the TREAT-NMD website: www.treat-nmd.eu/sma/care/family-guide/

The UK SMA Patient Registry

This leaflet describes the work of the Registry and how to sign up. A hard copy can be requested from SMA Support UK. It can also be downloaded from: www.treat-nmd.org.uk/registry

Glossary of Terms

Amino acid

The individual building blocks of **proteins**. There are 20 amino acids that are naturally incorporated into proteins. The specific order of the amino acids determines the structure and function of a protein.

Antibodies

Proteins made by the body to protect itself from "foreign" substances such as **viruses**.

Aspiration

Food or fluid that is breathed into the airway / lungs.

Atrophy

The wasting or shrinkage of a part of the body. SMA is called Spinal Muscular Atrophy because the lower **motor neurones** within the **spinal cord** degenerate which leads to the wasting of skeletal muscles.

Bulbar muscles

Muscles around the mouth and throat. When these muscles are affected swallowing and speaking become more difficult.

Cell

The basic structural and functional unit or building block of all known living organisms. A group of cells can work together to perform a common function to form a **tissue**. Cells come in many different forms such as **motor neurons** (a type of **nerve cell**), keratinocytes (main cell type of the skin), or erythrocytes (red blood cells). Humans have an estimated 100 trillion cells.

Central nervous system

The brain and **spinal cord**.

Chromosomes

Cylindrical shaped bundles of **DNA** in the **cell** nucleus (the core or centre of a cell). They consist of long threadlike strands of DNA coiled upon themselves many times. Humans have 46 chromosomes in each cell, they inherit 23 from their mother and 23 from their father to make 23 pairs. The only exceptions are sperm and egg cells which contain just 23 chromosomes each. **Genes** are located on the DNA which is packaged into the chromosome 'bundles'. The number of chromosomes can differ between species, for instance, chimpanzees possess 48 and dogs 78.

Clinical

Relating to the observation and treatment of patients rather than laboratory studies.

Clinical trial
A trial done on humans, usually to test a treatment or intervention or find out more about a disease.

Contracture
A tightness in the connective **tissue** and tendons around a joint that results from weakness and inability to move a joint through its full range of motion.

Deletion mutation
Genetic material (part of the **DNA**) missing from a chromosome or gene.

Diagnosis
Identifying a disease from its signs and symptoms or from its **genetic** cause. A **clinical** diagnosis is given when a doctor sees enough signs or symptoms to be confident that a person has the disease in question. In genetic diseases, a genetic diagnosis is given when a genetic test has been done and the fault in the **gene** that is known to cause the disease is found. Doctors who are experts in SMA can usually diagnose the condition with a high degree of accuracy from the clinical signs and symptoms alone, but genetic tests are usually recommended for all genetic diseases to increase certainty and to make sure the person has the "right" disease for any treatment.

Distal
Anatomical term of location meaning situated away from the centre of the body, towards the extremities. Distal muscles, such as those found in the hands and feet, are less affected by SMA compared to **proximal** muscles (those close to the centre of the body e.g. those in the hips, shoulders, and neck).

DNA
Deoxyribonucleic acid (DNA) is the molecule that contains the **genetic** instruction manual to build all known organisms. Short stretches of DNA sequence that code this information are known as **genes**. DNA is often compared to a set of blueprints, or a recipe, or a code, since it contains the instructions needed to construct other components of **cells**, such as **proteins**. DNA consists of nucleotides, the specific sequence of which determines the properties of the proteins made from the genes. Within cells DNA is organised into structures called **chromosomes**.

Electromyogram (EMG)
A test that assesses the electrical activity of the muscles and the nerves controlling the muscles. It is used to help diagnose **neuromuscular** disorders. There are two kinds of EMG in widespread use: intramuscular (needle) and surface EMG. To perform intramuscular EMG, a needle electrode, or a needle containing two fine-wire electrodes, is inserted through the skin into the muscle **tissue**. Intramuscular EMG may be considered too invasive or unnecessary in some cases and an electrode placed on the surface of the skin is used instead.

Enzyme

A **protein** which initiates, facilitates or speeds up a reaction. Almost all of the processes that occur in our body require enzymes. Examples include the digestion of food, the growth and building of **cells**, and all reactions involving transformation of energy.

Fasciculation

A small involuntary muscle twitch which can occur in any muscle in the body.

Gastrostomy tube (G Tube)

Feeding tube placed in the stomach in a surgical procedure. Sometimes referred to as a PEG (percutaneous endoscopic gastrostomy).

Gene

A stretch of **DNA** sequence that carries the information to produce a specific **protein**. Genes are the unit of **heredity** that are passed from one generation to the next. We usually possess two copies of each gene, one inherited from each of our parents. When genes are altered through **mutation**, this can affect the structure and function of the proteins that they produce, leading to disease.

Genetic counselling

Information and support provided by a specialist to people who have **genetic** conditions in their families or are concerned about a genetically transmitted condition. Genetic counselling helps families understand things like how the condition is passed on, what the chances are of children being affected, and which other family members may be at risk of carrying the affected **gene**.

Genetics

The study of **genes** and **inheritance**.

Heredity

The passing of traits through the **inheritance** of **genes** from one generation to the next.

Hypotonia

Low muscle tone.

Hypoventilation

Hypoventilation refers to a reduced rate and depth of breathing (too shallow or too slow) which leads to an increase of carbon dioxide (a waste gas) in the body.

Inheritance

The process by which an individual acquires characteristics from his or her parents or ancestors.

Motor neurone (or motor neuron)

The **nerve cells** that connect the brain and **spinal cord** to skeletal muscles allowing conscious movement. They act as a message delivery system: electrical signals originating in the brain are fired down the spinal cord along 'upper motor neurones' and on to skeletal muscles via the 'lower motor neurones'. Lower motor neurones are the main cell type affected by SMA.

Muscle biopsy

Removal of a small amount of muscle **tissue** for examination.

Mutation

A permanent change in the **DNA** sequence of a **gene** that can be inherited by subsequent generations. Dependent upon where a mutation occurs within the gene, it can have no effect on the **protein** produced, or can disturb the protein's function causing a **genetic** disorder such as SMA.

Nerve cell

Also called neurones, nerve cells are electrically excitable cells that allow the quick transmission of signals throughout the body. Different types of nerve cell make up the nervous system which functions to allow us to respond and react to our surroundings. For example, the brain sends a signal down the nerves to tell a muscle to contract.

Neuromuscular

Anything that relates to the nerves, muscles, or the nerve-muscle junction.

Orthopaedic

Relating to the musculoskeletal system: the body's muscles and skeleton, including the joints, ligaments, tendons, and nerves.

Orthoses (also orthosis and orthotics)

Devices or aids to prevent or assist movement of the spine or limbs or to provide support for joints and muscles. For example: splints, spinal jacket / brace, ankle-foot orthoses (AFOs), knee-foot orthoses (KFOs).

Protein

Consisting of chains of **amino acids** arranged in very specific orders, proteins are the building blocks of our bodies: fundamental components of living **cells** that are required for the structure, function, and regulation of cells, **tissues** and organs. The order of amino acids within a chain is determined by the **genetic** code (**DNA**), and different **genes** have the "instructions" for making different proteins. Proteins possess unique functions within a cell, and examples include **enzymes**, hormones, **antibodies** and the **survival motor neuron (SMN) protein**.

Proximal

Anatomical term of location meaning situated close to the centre of the body. Proximal muscles, such as those found in the hips, shoulders, and neck, are more affected by SMA than **distal** muscles, such as those in the hands and feet.

Rare Disease
The EU considers diseases to be rare when they affect not more than 5 per 10,000 persons in the European Union.[10]

Respiratory
Relating to breathing.

Scoliosis
Sideways curvature of the spine.

Sensory nerves
Nerve cells responsible for converting external stimuli, e.g. sound, light, smell, from the environment into internal signals. This is how we feel, see, and hear.

Spinal
Relating to the spine.

Spinal cord
The bundle of nervous **tissue**, which includes **nerve cells** and supporting **cells** and tracts, that extends out from the brain. The brain and spinal cord make up the **central nervous system** (CNS).

Survival motor neurone gene (SMN gene)
Mutations in the SMN **gene** are the cause of some forms of Spinal Muscular Atrophy. There are two types of SMN genes, ***SMN1*** and ***SMN2***.

Survival Motor Neuron (SMN) protein
Produced from both the ***SMN1*** and ***SMN2* genes**, the SMN protein is required for the survival of lower **motor neurons.** Complete absence of SMN protein is deadly to all different types of **cell**, but at low levels, the lower motor neurons appear to be the most severely affected cell type.

Survival Motor Neuron 1 (*SMN1*)
The **gene** that when mutated leads to the development of SMA. *SMN1* produces the full-length **SMN protein**, which we need a certain level of in order for our lower **motor neurons** to survive and thrive.

Survival Motor Neuron 2 (*SMN2*)
The **gene** that can modulate the severity of SMA because it is able to produce a small amount of functional full-length **SMN protein**. In people with a defect in the ***SMN1*** gene, this can be important because the more copies of *SMN2* that someone has, the more functional SMN protein they can produce. Patients with more severe forms of SMA (Type I/II) usually have fewer copies of the *SMN2* gene than patients with Type III SMA.

Symmetrical
Same on both sides of a central point.

Tissue

A collection of **cells** that work together to perform a common function. Organs are formed from multiple tissues working together with a common function.

Virus

Viruses consist of **genetic** materials surrounded by a protective coat of **protein**. They are capable of latching onto **cells** and getting inside them. Some viruses (like the cold virus or flu virus) cause people to become ill, but their ability to get inside cells also means certain viruses can be used to deliver treatments into the cell. Viruses are too small to be seen by the naked eye. They can't multiply on their own, so they have to invade a 'host' cell and take over its machinery in order to be able to make more virus particles. They can be made harmless by changing their genetic code.

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