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Spinal Muscular Atrophy Type B Awareness and Symptoms Prediction Using 3D Segmentation Process in MRI Images

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ABSTRACT

Spinal muscular atrophy (SMA) describes a group of disorders associated with spinal motor neuron loss. In this review we provide an update regarding the most common form of SMA, proximal or 5q SMA, and discuss the contemporary approach to diagnosis and treatment. Electromyography and muscle biopsy features of denervation were once the basis for diagnosis, but molecular testing for homozygous deletion or mutation of the SMN1 gene allows efficient and specific diagnosis. In combination with loss of SMN1, patients retain variable numbers of copies of a second similar gene, SMN2, which produce reduced levels of the survival motor neuron (SMN) protein that are insufficient for normal motor neuron function. Despite the fact that the understanding of how ubiquitous reduction of SMN protein leads to motor neuron loss remains incomplete, several promising therapeutics are now being tested in early phase clinical trials. This proposed model investigates the symptoms and scans readings from the initial MRI scan images of babies with mutation progress and SMN proteins formation benchmark values for this particular disorder SMA and further this segmented parameters are acquitted into the K-means clustering technique that predict the report with the disorder symptoms with MSE (mean square error) values that helps the babies in future to take prevention measures to overcome this problem.

Keywords : Spinal Muscular, K-Means Clustering, Neuron, Disorder Symptoms, MRI Images.

I. INTRODUCTION

Spinal Muscular Atrophy (SMA) is a progressive neuromuscular disorder. Usually, this condition is considered genetically induced with no known cure to date. Children are born with the condition and develop muscular weakness progressively as they grow. The weakness ultimately encompasses the whole muscular function rendering the limbs dysfunctional or paralyzed. Many children with SMA, if they do not have the weakness from the beginning, will start having the disease manifesting itself on the legs first and then the arms and, in due time, they will become quadriplegic and even more disabilities can follow including speech impairment. Assistive Technology support for people with such disabilities often requires identification of the best residual muscular function so that this can be utilized as a means of voluntary control.

Spinal muscular atrophy (SMA) refers to a group of inherited diseases of the motor nerves that cause muscle weakness and atrophy (wasting). The motor nerves arise from the spinal cord and control the

muscles that are used for activities such as breathing, crawling, walking, head and neck control, and swallowing. SMA is a rare disorder affecting approximately 1 out of every 10,000 individuals worldwide.

SMA affects muscles throughout the body. In the most common types, weakness in the legs is generally greater than in the arms. Sometimes feeding, swallowing, and respiratory function (e.g., breathing, coughing, and clearing secretions) can be affected. When the muscles used for breathing and coughing are affected and weakened, this can lead to an increased risk for pneumonia and other respiratory infections, as well as breathing difficulty during sleep. The brain's cognitive functions and the ability to feel objects and pain are not affected. People with SMA are generally grouped into one of four types (I, II, III, IV) based on their highest level of motor function or ability.

There are only a few reported muscle magnetic resonance imaging (MRI) studies of genetically-confirmed spinal muscular atrophy (SMA). The aim of this study was to search for the presence of selective muscle involvement and of disease progression by MRI in SMA 3b. MRI of proximal upper and lower extremities was performed on a 1.5-Tesla MR scanner using conventional T1 and T2 weighted axial images in twenty-two patients with genetically-confirmed SMA. Patients had detailed clinical examinations within one month of the radiological investigation. MRI revealed a selective involvement of muscles. Triceps, iliopsoas and quadriceps femoris were the most affected muscles. In contrast, biceps brachii, deltoid and gluteus maximus were well-preserved. Gracilis, short head of biceps femoris, semimembranosus, and Sartorius and adductor long us were also relatively spared. Slow, but definite progression with a predictable sequence and magnitude was documented. Median MRI grade was higher (worse) usually by one grade in the muscles at disease duration of >10 years compared to disease duration of ≤10 years. Longitudinal follow-up of four patients with two MRI studies eight years apart showed evidence of slow progression. Clinical-radiological comparison revealed a good correlation for most of the muscles. This muscle MRI study in a relatively large number of patients showed a specific pattern of involvement in SMA 3b and revealed that muscle MRI can be an objective and powerful tool in showing disease progression with implications for clinical trials.

Clinical Features Of SMA

Since only α motor neurons are lost progressively, only motor function is compromised and sensory neurons are unaffected. This loss of function leads to weakness and to progressive symmetrical atrophy of the proximal voluntary muscles of the legs, arms and, sometimes, the trunk, as the disease progresses. A number of unusual clinical features are observed in SMA. One of these is the distribution of muscle weakness, which is more compatible with a myopathic disorder than with a neurogenic disorder.12 Proximal muscles are more involved than distal muscles, legs are more affected than arms and arms are more affected than the face anddiaphragm.8, 12 In other words, muscle weakness and atrophy does not have a homogeneous distribution. The severity of muscle weakness is related to age at onset and children with the most severe form of the disease (Type I SMA) can appear normal at birth, but present muscle weakness few months later. Furthermore, the clinical course followed by SMA patients.

Who survive beyond childhood demonstrates that loss of muscle strength is normally most evident at diseases onset and that, after this, residual muscle power can remain stable for months or years.

II. LITERATURE SURVEY

One of the major challenges facing researchers engaged in the study of neurodegenerative and neuron development diseases is to explain why defects in ubiquitously expressed proteins have such a selective effect on the nervous system and its constituent cell types. Explaining the molecular mechanisms underlying the disease phenotypes has been hampered by their multifactorial nature and high incidence of sporadic cases. Alzheimer's disease, Parkinson's disease, and amyotrophic lateral sclerosis (ALS) are among the most common neurodegenerative diseases in humans and good examples of disorders that exemplify these challenges. Two less common neurological diseases which are monogenic in origin but are the result of defects in ubiquitously expressed proteins involved in housekeeping functions are Rett syndrome and 5q spinal muscular atrophy (SMA).

The former is characterized by mutations in the gene encoding methyl – CpG binding protein 2 (MeCP2), which likely regulates gene expression and chromatin structure; the latter by a deficit of the survival of motor neurons (SMN) protein, whose best characterized function is in snRNP biogenesis and pre-mRNA splicing. Yet, despite the apparent need for these proteins in all tissues, Rett syndrome, a prototypical neurodevelopment disorder, is characterized by a specific defect in maintaining proper function of post mitotic neurons in the forebrain, hippocampus, and brainstem, while SMA has a particularly profound effect on lower motor neurons.

This begs the question: do the proteins implicated in these diseases have functions in neurons other than their proposed housekeeping roles? Do they modulate downstream pathways specific to the tissues affected? Alternatively, are neuronal cells simply more susceptible than other tissues to genetic, biochemical, and environmental insults? Answering these questions using multifactorial diseases as paradigms poses an extra challenge precisely due to their complicated genetics. An attractive alternative would therefore be to address these issues in diseases with a simple Mendelian inheritance pattern. Spinal muscular atrophy provides researchers studying neurodegenerative diseases with this opportunity.

This review highlights why (1) SMA might be considered a prototypical neurodegenerative disease in which to address certain general questions facing the field, and (2) why a thorough understanding of this disorder might shed light on the mechanisms that have a specific effect on the development, health, survival, and causes of motor neuron degeneration, which ultimately leads to a disease phenotype. Proximal spinal muscular atrophy, commonly referred to as SMA, is a common autosomal recessive neuromuscular disease that affects the anterior horn cells of the spinal cord, resulting in atrophy of the proximal muscles of the limbs and trunk. There are numerous other forms of spinal muscular atrophy which share certain characteristics with proximal SMA; however, they are genetically distinct and often affect different subsets of neurons and muscle. They include autosomal dominant forms of the disease (Sambuughin et al., 1998; Van der Vleuten et al., 1998), X-linked forms (La Spada et al., 1991; Kobayashi et al., 1995), recessive forms that affect the distal muscles (Viollet et al., 2002), and a severe form of SMA (SMARD) with respiratory distress (Grohmann et al., 2001) (Table 1).

After cystic fibrosis, SMA is the most common autosomal recessive disorder in humans, with a carrier frequency of approximately 1 in 35 and therefore an incidence of 1 in 6000 in the human population. It is also the most common genetic cause of infant mortality. Despite the high incidence of the disease in the human population, SMA has gained relatively little attention among researchers studying neurodegenerative diseases. However, it has a fascinating biology, which includes two major players: the highly homologous SMN1 and SMN2 genes.

A splicing defect in SMN2 is a key factor in causing the disease phenotype, while the SMN protein, expressed by both genes, is very likely multifunctional. However, low levels of the protein have a particularly detrimental effect on one tissue type, the lower motor neurons. One of SMN's functions is essential to cell survival. These characteristics, coupled with a relatively simple Mendelian inheritance pattern, make a compelling case that study of this disorder in more detail will provide a prototype that might shed considerable light on motor neuron biology and disease. Such studies may eventually lead to a better understanding of SMA and other similar diseases and may accelerate progress toward rational therapeutics.

III. METHODOLOGY

Magnetic Resonance Imaging is a fast and high resolution technique that is commonly used to investigate the anatomy and physiology of the body in health and disease. High resolution imaging of the important heart elements has become possible through Multi-slice MRI technology. There has been a very large body of studies using MRI scans to segment body parts for research purposes and also to increase the efficiency of the clinician's work.

There are **three major parts** of the proposed approach to segment 3-D MRI SMA Data with minimum user interaction.

In the first step, a pre-processing operation is employed to remove noise from individual slices.

Second, the user is required to provide soft constraints to incorporate information about spatial distance of regions to a reference point.

In the third step, the smoothness term can be adjusted based on the input data. For example, if some labels cannot be adjacent to each other, then a higher penalty is applied for the neighbouring pixels that are assigned those labels for the given neighbourhood system. Finally, the 3-D graph cut structure is formed and segmentation is performed.

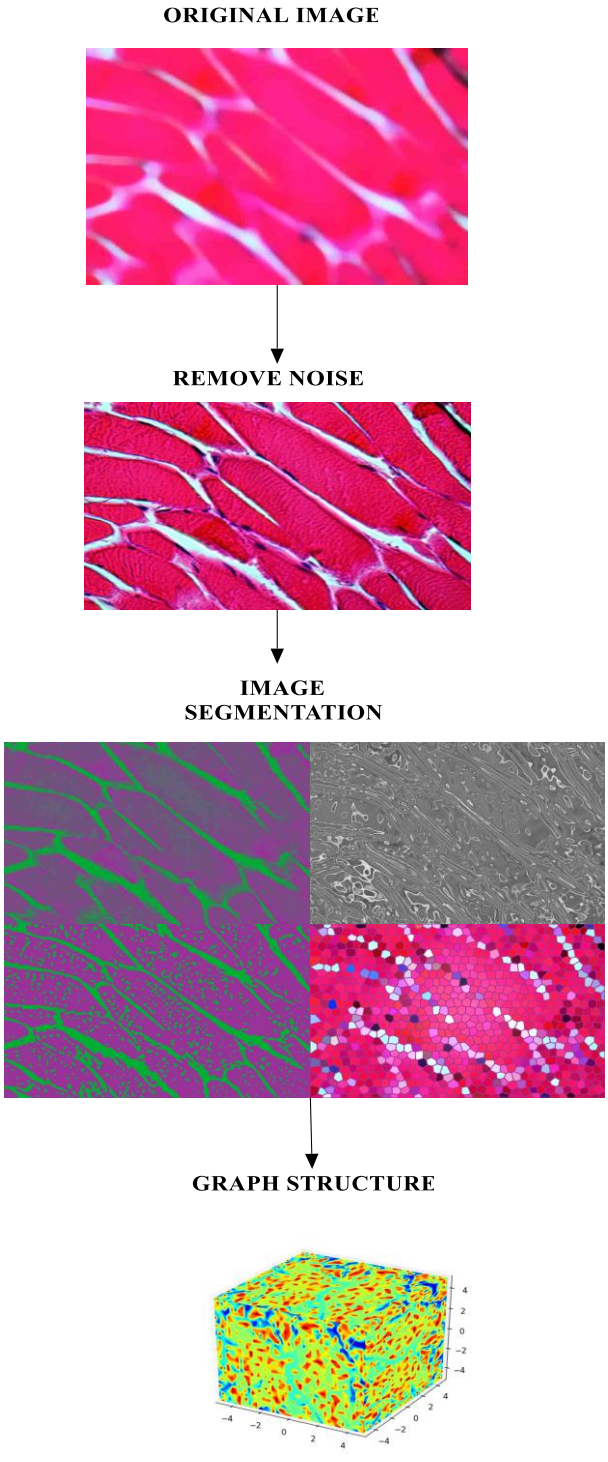


Fig.1. Work Flow

STEPS INVOLVED:

- Respondents were invited by Cure SMA to participate in the survey via email containing a link to the survey.
- Responses were collected, tallied and analyzed by Silicon Valley Research Group.
- No PHI was collected from survey respondents through the survey or by any other means.
- The survey was submitted to the IRB for approval prior to data collection and is IRB approved and compliant.
- The total sample size for the study was 298 completed responses, yielding a margin of error of 5.68%.
- A possible explanation for the consistency in most tolerable and least tolerable risks across all treatments is that SMA sufferers and their caregivers consider any of the benefits of treatments presented as equally important, i.e. they are not trading off risks for different treatments.
- Survey respondents also appeared to weigh risks against their probabilities of occurrence. Consistently, high probabilities of occurrence made a risk less tolerable.

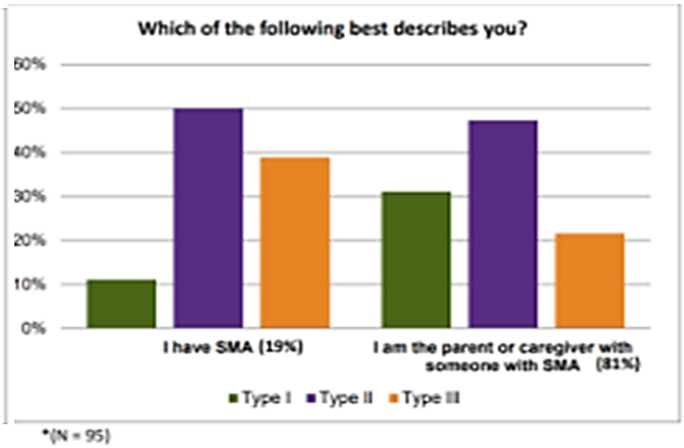
IV. RESULT AND DISCUSSIONS

Both medical follow-up and palliative care are important throughout SMA patients' entire lives. This care includes respiratory and nutritional support, and orthopaedic and physiotherapeutic care to avoid postural disorders.

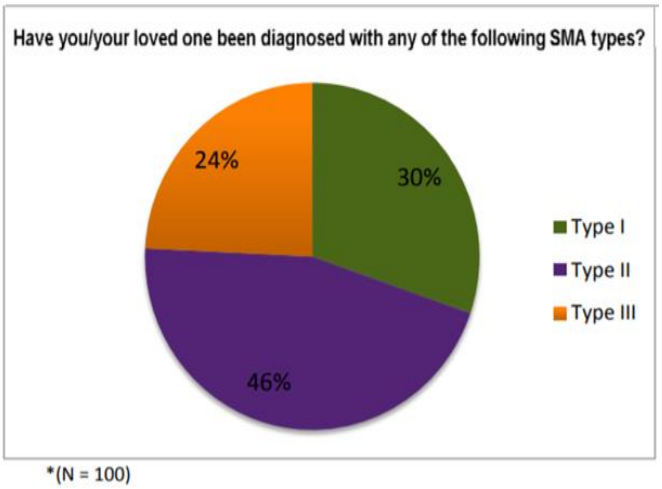
In addition to this we can cite pharmacological treatments that are still being studied, both employing new drugs and drugs that are already well-known. It cannot be expected that the pharmacological treatments currently being studied can recover motor neurons or muscle cells that have already been lost to atrophy.

Rather, the objective is to retard the progress of the disease and improve residual muscle function. Sadly, paralysis can be halted, but it cannot be reversed. However, through a combination of medical care and rehabilitation, many patients with SMA can have fulfilling and productive lives and often have normal life expectancy.

Finally, there is one other point it is important to emphasize, which is that since SMA is a recessive genetic disorder, genetic counselling is an essential component forthe families of these patients. Through genetic counselling,



Graph.1. Efficiency Analysis



Graph.2. Diagnosed SMA

V. CONCLUSION

It is obvious that significant challenges remain toward an eventual treatment for SMA. These stem from the many unanswered questions this review has raised. Readers may come away thinking that SMA research is still in its infancy. However, it should be noted that the tools to answer these questions, i.e., an excellent set of animal models and a basic understanding of the disease, already exist. It should also be reiterated that there are a number of compelling reasons investigators outside the field may find problems in SMA an attractive challenge to take on; for example, SMA is a neurodegenerative disease in which a defect in a ubiquitously expressed protein affects a very specific tissue type. The genetics of the disease are also relatively simple: SMA is autosomal recessive and involves a loss of function in an essential protein in certain cells. In essence, SMA results from an insufficient amount of the SMN protein.

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