THE COURSE OF SMA IN THE HOME CARE PATIENT: 3 CASE STUDIES.

Bruce Estrem[1], Carrie A. McKenzie[1], Carrie L. Priztl[1], Paul Kubic[2]; [1]Pediatric Home Service, Roseville, MN; [2]Pediatric Pulmonology, Children's Hospitals and Clinics, St. Paul, MN

Introduction: Spinal Muscular Atrophy (SMA) is the most common genetic cause of infant mortality. Mutation of the SMN-1 gene is present in 1:35 adults, resulting in a population of 1:6,000 suffering from the disease. After cystic fibrosis, SMA is the most common autosomal recessive disorder in humans. Researchers have identified 4 primary classifications of SMA; Type I (acute infantile SMA, Werdig-Hoffman disease) typically found in infants. Type II (intermediate SMA) is usually identified 6 – 12 months of age. Type III (chronic SMA, Kugelberg-Welander disease) diagnosed after 18 months of age. Type IV presents in the late teens to mid 20's years of age. Bulbospinal muscular atrophy (Kennedy Syndrome) is usually found in adults, 20 – 40 years of age is sometimes referred to as SMA Type V. Diagnosis and treatment for SMA is a combination of patient symptoms, family history and genetic testing that will show a mutation in a single gene; Survival of Motor Neuron 1 (SMN-1). Performing an electromyelogram may reveal damaged nerve impulses. Muscle biopsy is definitive in the diagnosis of childhood SMA to verify atrophy of muscle fibers. Currently, the plan of care for SMA is limited to symptom management since there is no known treatment to reverse the mutation of the SMN-1 gene. Presentation; We will present 3 case studies that will highlight different treatment pathways: A family that chose not to be aggressive with treating the symptoms (which resulted in a short life span for their child), a child supported through non-invasive mechanical ventilation (Bilevel to mask), and a young adult supported invasively with a ventilator through a tracheostomy. Discussion; Advances in health care have allowed SMA patients to be more aggressive with medical intervention. Although neuromuscular functions of the body deteriorate, cognitive capabilities are not affected. Therefore ethics, culture, and heritage all play a large part in the integration of mechanical ventilation, aggressive pulmonary hygiene and nutritional support. When a patient or family chooses medical intervention in the form of mechanical support, there are two distinct areas where improvements can be made. First, domestic availability of infant and pediatric sized non-invasive interfaces (masks). Second, increased portability through small, lightweight, battery operated ventilators and bi-level devices with affordable battery options.