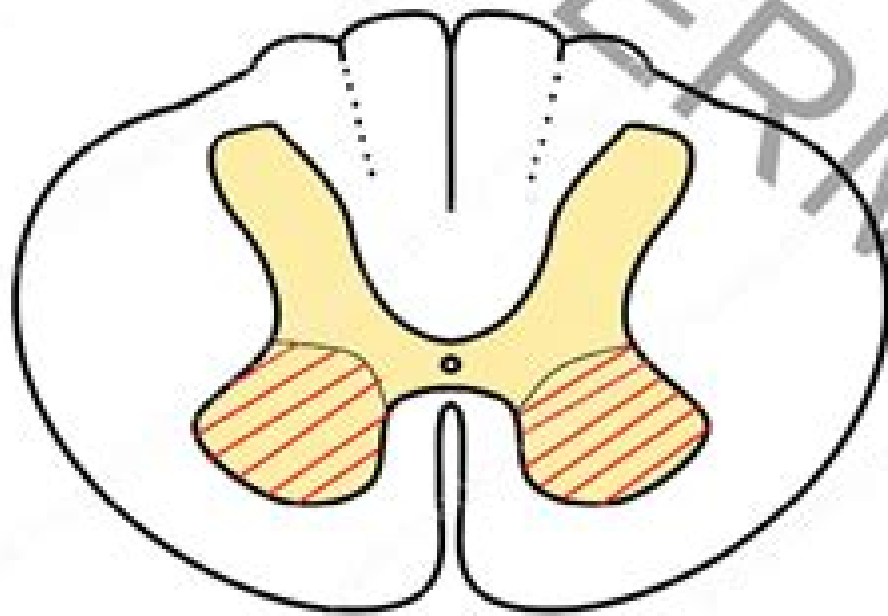


# Spinal Muscular Atrophy (SMA)

Autosomal recessive mutation in **SMN**

SMA type 1 = Werdnig-Hoffmann disease



Congenital degeneration of  
anterior horn cells (**LMN** death)



- Floppy baby
- Symmetrical weakness
- Fasciculations
- Respiratory failure

# It S One Of Them Living With Spinal Muscular Atrophy

**Michael Hoelscher**



## **It S One Of Them Living With Spinal Muscular Atrophy:**

It's One of Them! Grace Saunders,2014-12-10 Grace Saunders and her brother Ben were both born with the genetic disorder spinal muscular atrophy type 2 3 SMA From her happy childhood growing up in Hertfordshire enduring painful operations to becoming a mom and a wife living an independent life in Coventry Her extraordinary story of strength not only coping with a severe physical disability but being victim to domestic violence Graces story also involves drugs murder the police investigation that was Operation Ore and the death of her brother aged thirty This is a womans struggle for a normal life independence and remarkably having a healthy baby girl when she was told she would never be able to have children She coped many years being a single mother with the help of her family and personal care assistants She writes about her education care and funny stories about the world of Internet dating She tells her story with honesty humor and heartfelt emotion Grace shares the highs and lows she has faced and has come out the other side although a little bruised stronger than ever and still smiling Graces attitude to the life she has been dealt with is as she says Its One of Them **Spinal Muscular Atrophy** Charlotte J Sumner,Sergey Paushkin,Chien-Ping Ko,2016-10-24 Spinal Muscular Atrophy Disease Mechanisms and Therapy provides the latest information on a condition that is characterized by motoneuron loss and muscle atrophy and is the leading genetic cause of infant mortality Since the identification of the gene responsible for SMA in 1995 there have been important advances in the basic understanding of disease mechanisms and in therapeutic development This book provides a comprehensive accounting of recent advances in basic and clinical research that covers SMA clinical features and standards of care multifaceted aspects of SMN protein functions and SMA disease pathology various animal models and biomarkers as well as current therapeutic development This title is ideal for graduate students postdocs and principal investigators who are already in the SMA field and need to keep updated on recent findings and approaches and for those who are new to or would like to join the field Likewise users will find an excellent source of reading for biotech pharma scientists clinical researchers and practitioners regulators and patients and their advocacy organizations Furthermore this book is a handy reference for researchers and clinicians who may want to apply the research strategies and therapeutic approaches in SMA to other rare diseases Provides comprehensive up to date reviews by leading investigators on diverse topics of SMA including clinical features and patient care SMN genetics and protein functions animal models disease pathology and mechanisms biomarkers current therapeutic development and the role of non profit organizations in therapeutic development Written to bridge multiple disciplines and promote better communications among basic scientists clinical researchers and health care providers on the latest developments in SMA Includes outstanding questions and perspectives for future investigations and key references for additional detailed study The Encyclopedia of Genetic Disorders and Birth Defects James Wynbrandt,Mark D. Ludman,2010-05-12 Presents information on congenital birth defects giving the name known or suspect cause and brief description as well as available treatments where applicable *American*

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