ABOUT UBA5 DISEASE

WHAT CAUSES UBA5 DISEASE?

The Ubiquitin-like modifier-activity enzyme (UBA5) gene is universal and criticall as it contains instructions necessary for maintaining cellular breakdown and function. This disease is caused when the UBA5 gene is mutated, resulting in malfunction.

HOW RARE IS THIS DISEASE?

There are only about 30 documented UBA5 disease cases in the world, but it spans across America, Europe, Australia and Asia. Because of how ultra-rare the disease is, there is a significant lack of research and attention given to those affected.

WHAT ARE THE SYMPTOMS?

MOTOR CONTROL ISSUES

Hypotonia (body floppiness)
Spasticity (body stiffness)
Dystonia (involuntary muscle contraction)

COGNITIVE ISSUES

Seizures (some are uncontrollable)
Intellectual disability
Brain structure abnormalities

GROWTH ISSUES

Microcephaly (poor health growth)
Failure to thrive

OTHER MEDICAL ISSUES

Respiratory complications
Gastroesophageal reflux
Vision impairment

WHAT IS THE EFFECT OF UBA5 ON QUALITY OF LIFE?

Most children with UBA5 disease cannot sit, walk, or talk. They are often get food via a feeding tube due to their inability to swallow safely. They require 24/7 care and multiple medicines along with daily therapies to maintain their health and survival.

HOW CAN GENE THERAPY HELP?

Gene therapy offers hope for those affected by UBA5. This medical innovation works by replacing or adding a new gene to treat diseases caused by a faulty gene. The advancement and success of gene therapy in other disease areas makes it possible for UBA5, but it is continuing to be developed and is a costly process.