WHAT ARE SPATA5 AND SPATA5L1 RELATED DISORDERS?

SPATA5 (also called AFG2A) and SPATA5L1 (also called AFG2B) are two genes found in the body. When these genes become mutated, it results in SPATA5 and SPATA5L1 Related Disorders.

There are approximately 150 diagnosed world-wide with these conditions.

The function of these genes is not fully understood and no treatment currently exists. Mutations in either gene cause the same set of symptoms.

It's important to note that every case is different, and not everyone affected will experience all symptoms, and symptoms may be experienced at various degrees of severity. There may be other symptoms present related to these disorders, but these are the most common.

Learn more about SPATA5 and SPATA5L1 Related Disorders and the work The SPATA Foundation is doing at www.SPATAfoundation.org.

MOST COMMON SYMPTOMS

- Hearing Loss
- Developmental Delay
- Physical/Intellectual Disability
- Epilepsy
- Feeding Difficulties
- Hypotonia
- Spasticity
- Dystonia
- Chorea
- Ataxia
- Vision Impairment
- Abnormal MRI
- Microcephaly
- Neurostorming