

## Gene

**HEXA**

## Associated Diseases

Tay-sachs Disease

## Phenotype

### Apathy

#### **Psychomotor deterioration**

Loss of previously present mental and motor abilities.

#### **Pallor**

Abnormally pale skin.

#### **Generalized hypotonia**

Generalized muscular hypotonia (abnormally low muscle tone).

#### **Seizure**

A seizure is an intermittent abnormality of nervous system physiology characterised by a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain.

#### **Cherry red spot of the macula**

Pallor of the perifoveal macula of the retina with appearance of a small circular reddish choroid shape as seen through the fovea centralis due to relative transparency of the macula.

#### **Exaggerated startle response**

An exaggerated startle reaction in response to a sudden unexpected visual or acoustic stimulus, or a quick movement near the face.

#### **Blindness**

Blindness is the condition of lacking visual perception defined as visual perception below 3/60 and/or a visual field of no greater than 10 degrees in radius around central fixation.

#### **Hypotonia**

Hypotonia is an abnormally low muscle tone (the amount of tension or resistance to movement in a muscle). Even when relaxed, muscles have a continuous and passive partial contraction which provides some resistance to passive stretching. Hypotonia thus manifests as diminished resistance to passive stretching. Hypotonia is not the same as muscle weakness, although the two conditions can co-exist.

#### **Autosomal recessive inheritance**

A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

**Hypertonia**

A condition in which there is increased muscle tone so that arms or legs, for example, are stiff and difficult to move.

**Poor head control**

Difficulty to maintain correct position of the head while standing or sitting.

**GM2-ganglioside accumulation**

Cellular accumulation of GM2 gangliosides.

**Aspiration**

Inspiration of a foreign object into the airway.

**Dementia**

A loss of global cognitive ability of sufficient amount to interfere with normal social or occupational function. Dementia represents a loss of previously present cognitive abilities, generally in adults, and can affect memory, thinking, language, judgment, and behavior.