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ANGELMAN SYNDROME & HOMOEOPATHY

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Abstract:
Angelman syndrome is a genetic disease condition that causes developmental delays, speech and balance problems, intellectual disability and seizures[1]. The diagnosis is based on history, unusual movements and characteristic facial appearance. There is no current treatment available. Symptomatic treatment according to the symptoms like anticonvulsants for seizures.

Introduction

Angelman syndrome (AS) is characterized by severe developmental delay or intellectual disability, severe speech impairment, gait ataxia and/or tremulousness of the limbs, and unique behavior with an apparent happy demeanor that includes frequent laughing, smiling, and excitability. Microcephaly and seizures are also common. Developmental delays are first noted at around age six months; however, the unique clinical features of AS do not become manifest until after age one year[2].

Angelman syndrome is due to a lack of function of part of chromosome 15, typically due to a new mutation rather than one inherited from a person's parents. It is due to a deletion or mutation of the UBE3A gene on that chromosome. It affects 1 in 12,000 to 20,000 people.Males and females are affected with equal frequency[3].

It is named after British pediatricianHarry Angelman, who first described the syndrome in 1965[4].

Clinical Features[5]

The Symptoms occur at relative frequency and vary from person to person. Common symptoms includes

Developmental delay,

- Speech impairment,
- Movement or balance disorder,
- Behavioraldisorders

- Delayed, disproportionate growth in head circumference,
- Seizures, onset usually less than 3 years of age
- Abnormal EEG, characteristic pattern with large amplitude slow-spike waves
- Strabismus
- Hypopigmented skin and eyes
- Tongue thrusting; suck/swallowing disorders
- Hyperactive tendon reflexes

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- Feeding problems during infancy
- Uplifted, flexed arms during walking
- Prominent mandible
- Increased sensitivity to heat
- Wide mouth, wide-spaced teeth
- Sleep disturbance
- Frequent drooling, protruding tongue
- Attraction to/fascination with water
- Excessive chewing/mouthing behaviors
- Flat back of head
- Smooth palms

Diagnosis[5]

The diagnosis of Angelman syndrome is based on

- A history of delayed motor milestones and then later a delay in generaldevelopment, especially of speech
- Unusual movements including fine tremors, jerky limb movements, hand flapping and a wide-based, stiff-legged gait.
- Characteristic facial appearance (but not in all cases).
- A history of epilepsy and an abnormal EEG tracing.
- A happy disposition with frequent laughter
- A deletion or inactivity on chromosome 15 by array comparative genomichybridization (aCGH) or by BACs-on-Beads technology.

Diagnostic criteria for the disorder were initially established in 1995 in collaboration with the Angelman syndrome Foundation (US);[6] these criteria underwent revision in 2005.[7]

Seizures are a consequence, but so is excessive laughter[8], which is a major hindrance to early diagnosis.

Differential diagnosis

Other conditions that can appear similar include:[9,10]

- Autism spectrum
- Cerebral palsy
- Rett syndrome
- Mowat Wilson syndrome
- Adenylosuccinate lyase deficiency

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- Pitt Hopkins syndrome
- Phelan McDermid syndrome
- Prader Willi syndrome

Treatment

There is currently no cure available. The epilepsy can be controlled by the use of one or more types of anticonvulsant medications.

Occupational therapies like such as finger isolation, motor planning, hand-eye coordination, spatial awareness, and refining gestures[11].

Homoeopathic Management[12]

Homoeopathy provides a promising result in such patients. Medicines given on individualistic approach based on totality of the symptoms produces wonderful effect in the treatment of symptoms of Angelman Syndrome. Common Medicines indicated in such cases includes:

- Calcareacarbonica
- Bufo rana
- Cuprum Metallicum
- OEnanthecrocata
- Artemisia vulgaris
- Kali bromatum
- Silicea
- Nux vomica
- Plumbum
- Cicutavirosa
- Sulphur
- Hyoscyamus
- Belladonna
- Causticum

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