Misdiagnosis

Individuals with Cohen Syndrome have been misdiagnosed with the following disorders:

- Prader- Willi Syndrome
- Angelman Syndrome
- Bardet-Biedl Syndrome
- Williams Syndrome
- Mirhosseini-Holmes-Walton Syndrome

How to Help

Your tax-deductible donation will be used to educate professionals and families about Cohen Syndrome with the goal of earlier diagnosis. Donations can be accepted online at our website at:

http://cohen-syndrome.org

Or

Please support Cohen Syndrome Families with sending your tax deductible donation to

DDC Clinic for Special Needs Children:
14567 Madison Road, Middlefield, OH 44062

The Many Faces of Cohen Syndrome

Cohen Syndrome Association
14567 Madison Road, Middlefield, OH 44062

Contact us at
info@cohen-syndrome.org

Cohen Syndrome Association

We A.R.E. Cohen Syndrome

We **A**dvocate, **R**esearch **&** **E**ducate families

And professionals about Cohen Syndrome

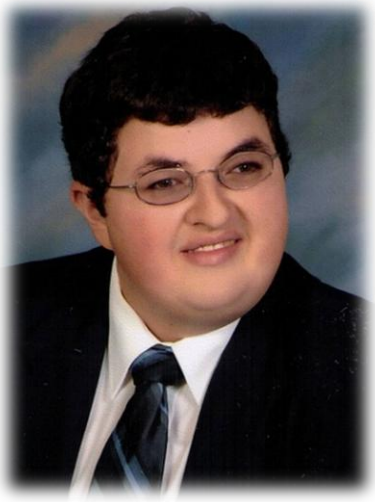
Overview

Cohen Syndrome is a rare genetic disorder associated with mutations present at COH1 and is often undiagnosed for many years. Many times the diagnosis is found as a result of emerging clinical symptoms. The Cohen Syndrome Association was founded by parents to raise awareness of this disease with the goal of educating parents and professionals to assure earlier diagnosis and medical interventions. Globally the total number of Cohen Syndrome children is estimated to be less than one thousand; however, it is believed to be undiagnosed or misdiagnosed in many cases.

Clinical symptoms

Symptoms often go unnoticed during the first few months of life until parents notice their children are not meeting developmental benchmarks. Clinical symptoms of Cohen Syndrome are:

- Failure to thrive in infancy
- High myopia and/or retinal dystrophy
- Microcephaly
- Developmental delay
- Joint laxity
- Narrow hands and feet and small stature
- Friendly disposition
- Neutropenia
- Characteristic facial features



Early diagnosis and daily life

Early diagnosis of Cohen Syndrome can make a positive impact as this enables medical and therapeutic interventions to ensure children thrive.

Intensive therapies such as O.T., P.T. and Speech are critical to ensure maximum development of gross/fine motor functions.

Intellectual disability impacts all cases with a wide variance in degree of impairment.

Vision issues impact daily life with progressive high myopia, night blindness, and retinal dystrophy in some cases. Annual ophthalmologic exams are needed to access changes in vision acuity.

Speech is affected in various degrees - late emerging speech, poor articulation, and total apraxia (nonverbal) in some cases.

Chronic Neutropenia can be attributed to frequent illness for some children leading to dental issues and mouth sores. Neupogen injections can often reduce recurrent infections and improve overall health.

Cohen Syndrome children are known to have a very friendly disposition and a positive outlook.