DeSanto-Shinawi

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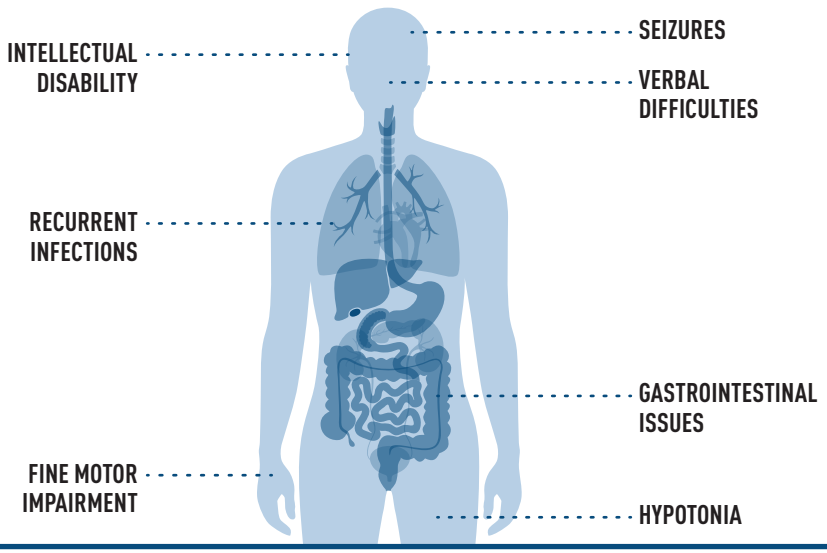
DeSanto-Shinawi Syndrome (DESSH) is a rare neurodevelopmental disorder that may affect different parts of the body. It is characterized by a mutation of the WAC gene. The mutation causes the gene to lose function and fail to produce the WAC protein in all cells.

Many patients with DESSH have cognitive abilities that range from mild to moderate and to date, it seems while there are many similarities, no two cases are the same. **At present, there is no overarching medical study and summary of the issues associated with DESSH.*** This resource is based on symptoms reported by those caring for people with DESSH, and anecdotal medical advice. It's intended to provide an overview of what patients have experienced to better inform medical professionals and caregivers, until science catches up on this recently identified rare syndrome.

Individuals with DESSH may begin to miss milestones as early as infancy. It was discovered in 2015, and is classified as a rare disease with fewer than 200 cases diagnosed globally. There is no cure, but therapeutic and medical interventions are necessary to improve patients' functioning and abilities. DeSanto-Shinawi Syndrome is characterized by global developmental delay, hypotonia, behavioral problems, intellectual disability, feeding difficulty, and seizures, among other symptoms. It particularly affects speech - and individuals can find it hard to communicate. The long-term impact of DESSH can vary between individuals. Individuals with DESSH are capable of living long full-filling lives.



COMMONLY SEEN SYMPTOMS

Symptoms that have been noticed in many subjects with DESSH also include:

- global developmental delay
- hypotonia
- verbal difficulties (including dysarthria + being non-verbal)
- apraxia
- immunodeficiency (including hypogammaglobulinemia)
- frequent recurrent infections (including respiratory + renal)
- respiratory issues (asthma + poor ability to clear lung passages)
- gastrointestinal issues + abnormalities (including GERD)
- severe constipation
- ocular abnormalities (strabismus, cortical impairment + astigmatism)
- feeding difficulty
- hearing issues (including loss + deafness)
- cardiovascular issues (including congenital heart disease, palpitations)
- cardiac anomalies
- seizures
- early onset puberty
- growth hormone deficiency
- kidney abnormalities
- pelvic abnormalities
- limb abnormalities
- bladder + bowel issues (including nocturnal enuresis + encopresis)
- dental issues (including hypoplasia)
- hirsutism
- sleep disturbances
- weight problems (including dyslipidemia)
- brain + congenital anomalies
- brain atrophy
- keratosis pilaris
- thin corpus callosum
- disorders affecting white matter
- fine motor impairment
- limited range of motion
- scoliosis
- symptoms associated with ADHD, ASD, Anxiety + SPD

Appearance Markers:

There are appearance markers that have been noted in scientific literature. Almost all patients have dysmorphic features, some of which can include: microcephaly, a broad or prominent forehead, brachial clefts, synophrys and/or bushy eyebrows, deep-set eyes, depressed nasal bridge, short nose, bulbous nasal tip, flaring nostrils, preauricular pits, posteriorly rotated and/or simple ears, large tongue, large teeth, thin upper lip, downturned mouth, broad mouth, broad chin, inverted nipples, coccygeal dimple, malformed toenails, coarse face, short fingers, short stature, a square shaped face, long palpebral fissures, and brachycephaly.

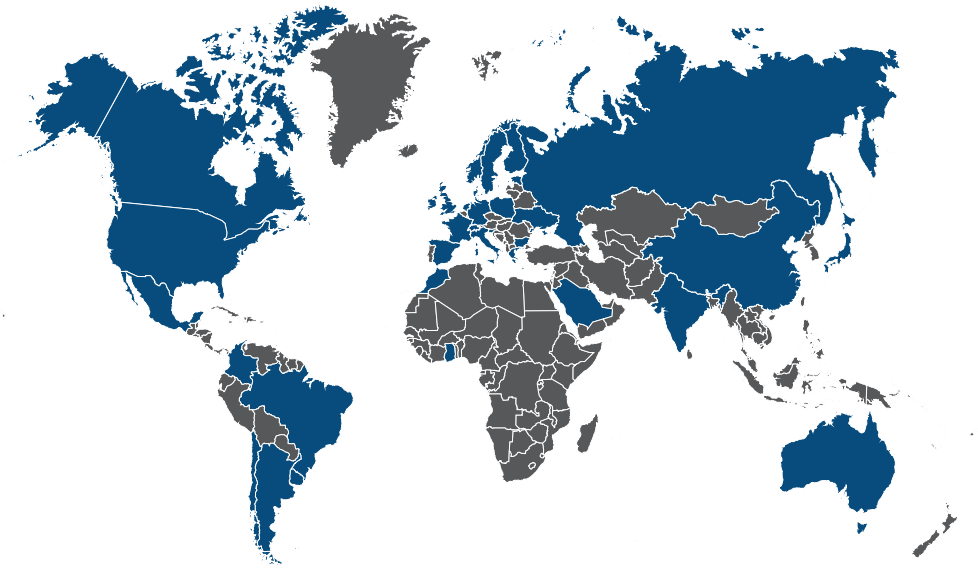
Behaviors:

Many with DESSH can also experience symptoms of, or be diagnosed with, neurodivergent conditions, secondary to DESSH. Diagnosis to date include Autism Spectrum Disorder (ASD), Attention Deficit Hyperactivity Disorder (ADHD), Anxiety, Bipolar, Obsessive Compulsive Disorder, Intermittent Explosive Disorder, and Mixed Receptive-Expressive Language disorder.

Diagnosis:

DESSH is definitively diagnosed using Whole Exome Sequencing (WES), a type of genetic test. Prior to an official diagnosis, many are left with no formal diagnosis, a diagnosis of Autism or a diagnosis of Cerebral Palsy.

Countries with Registered DESSH Patients ▸
(AS OF 2023)



Treatment:

While there is no treatment for DeSanto-Shinawi Syndrome, **each of the symptoms associated with the syndrome may be individually managed**. Hypotonia, for instance, reports excellent results with physical therapy, particularly if started early and at a maintained consistency. Speech-Language Pathology, Feeding Therapy, Occupational Therapy and Behavioral Therapy are also frequently needed. Additionally, Hippotherapy (equine therapy) may also be used. Stimulating environments, sensory tools, behavioral interventions, medications, medical devices and support systems are often recommended. Children with DESSH usually require additional support in their learning environment. The type of education depends on their individual learning needs.

The care team for a person with DESSH may consist of the following specialists:

- immunology
- behavioral pediatrics
- gastroenterology
- genetics
- neurology
- podiatry
- ophthalmology
- audiology
- pulmonology
- cardiology
- plastic surgery
- orthopedics
- prosthetics
- psychiatry
- special needs dentist
- nephrology
- endocrinology
- urology

***Recent references:** Pasquali D, et al., Patients with DeSanto-Shinawi Syndrome: Further extension of phenotype from Italy. Am J Med Genet. 191A:823-830. 2023. DOI: 10.1002/Ajmer.a.63061.