



NEUROGENE



INVITAE

Sponsored, no-charge genetic testing and counseling for patients suspected of having a lysosomal storage disease (LSD)

What is Aspartylglucosaminuria (AGU)?

- Aspartylglucosaminuria (AGU) is a rare neurodegenerative LSD caused by deficient enzyme activity due to pathogenic variants in both copies of the *AGA* gene.
- Reduced activity in the aspartylglucosaminidase (AGA) enzyme leads to the build-up of toxic chemicals that affect many organs of the body including the brain and spinal cord. AGU affects both males and females.
- Early symptoms of AGU usually appear by the age of 12 to 15 months and include developmental delay, specifically lack of speech and clumsy walking, and chronic ear infections. New signs and symptoms of the disorder develop and progress over time.

Developmental delay is an early observed symptom of AGU. Common misdiagnoses for AGU may include autism and attention deficit hyperactivity disorder (ADHD) as awareness of the condition is very low.

How AGU Affects the Body

AGU affects children both cognitively and physically. Aspects of the disease may include:

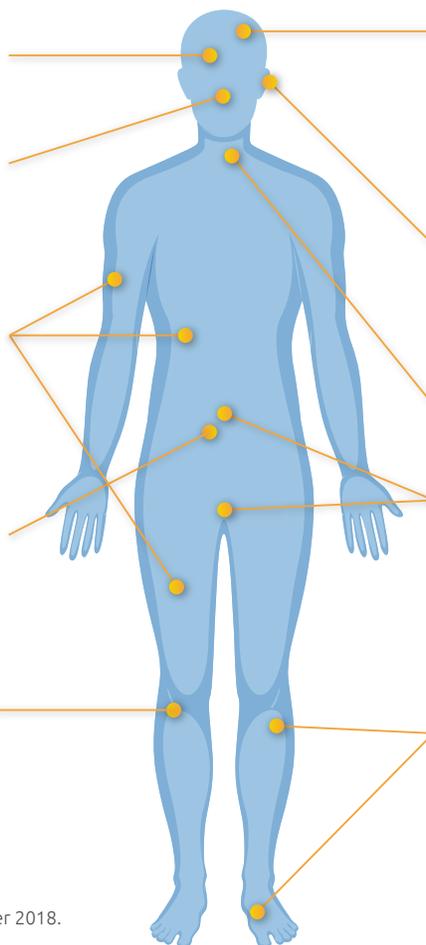
progressive thickening of facial features:
widely-spaced eyes, small ears, full lips,
acne, broad nose

dental problems, tooth decay, enlarged
tongue and tonsils

rib and breast plate
deformities, scoliosis and
other bone deformities
of the long bones
(i.e. humerus or femur)

gastrointestinal (GI) problems,
including diarrhea or constipation

unusually large range
of motion in joints
(hypermobility)



developmental delay, speech delay,
progressive intellectual decline,
confusion with simple instructions,
anxiety, behavioral issues, sleep
problems, seizures (> 20 years old)

chronic ear infections in early childhood
often starting (< 5 years old) requiring
multiple ear tube placements

frequent respiratory infections that may
lead to the removal of tonsils

hernias around belly button or groin

walking difficulties: including
rolling instep, toe walking*,
and "knock knees"



Why Test?

Genetic testing and counseling may help to:

- Identify risk of disease for patients and their family members
- Shorten the time to diagnosis and prevent misdiagnoses
- Seek appropriate care and treatment
- Determine if patients are eligible to participate in clinical trials
- Provide families with information about support resources and patient advocacy organizations

About Neurogene

The Invitae Detect LSDs is co-sponsored by Neurogene Inc., which is focused on developing life-changing genetic medicines for patients and their families affected by rare, devastating neurological diseases. We partner with leading academic researchers, patient advocacy organizations and caregivers to bring therapies to patients that address the underlying genetic cause of a broad spectrum of neurological diseases where no effective treatment options exist today. Our lead programs use AAV-based gene therapy technology to deliver a normal gene to patients with a dysfunctional gene. Neurogene is also investing in novel technology to develop treatments for diseases not well served by gene therapy. For more information, visit www.neurogene.com.



Please visit
invitae.com/en/detectLSDs
to order a no-charge genetic test.

While third parties and commercial organizations may provide financial support for this program, tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing and counseling are available in the US and Canada. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any other products or services from Invitae or from third parties or commercial organizations.

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