



G-Autism

Hereditary Autism Risk Screening 물

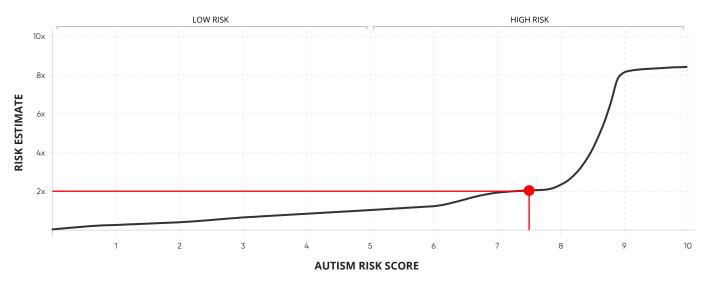






FOR		TEST INFO	
User	Nguyen Van A	Genetica ID	QWEKSU8UY6T
DOB	10 October 1990	Specimen	Saliva
Gender	M	Received	N/A

RESULT



- AUTISM RISK SCORE: 7.5/10
- **RISK ESTIMATE**: The individual's risk of autism is 2.01x.
- 3123 variants associated with Autism Spectrum Disorder were found in the individual's genome. Among them are:
 - 0 protective variants
- 0 pathogenic variants
- 13 harmful variants

Gene	Location	Value	Effect	Status	Impact
CNTNAP2	146792514	AT NM_014141.6(CNTNAP2):c .208+18133	Medium	Harmful	Possibly higher risk of autism due to change in nervous system development gene
ARID1B	157347549	CC NC_000006.12:g.1573475 49	Medium	Harmful	Susceptible to mild mental impairment, linked to harmful mutation in the ARID1B gene
CYFIP1	22896157	CC NC_000015.10:g.2289615 7	Medium	Harmful	Increased risk of having hallucinations that's tied to a schizophrenia-related gene
LINC01104	48968728	TG NC_000014.9:g.48968728	High	Harmful	Susceptible to social communication difficulties, linked to an increased risk mutation of autism
WNT2	117279924	AA NC_000007.14:g.1172799 24	High	Harmful	Susceptible to autism-associated language delay in uttering his first complete sentence

(To view the full list of ASD associated variants, download the Genetica mobile app from App Store or Google Play.)



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PATHOGENIC MUTATION

• NEGATIVE: 0 pathogenic mutations detected in the individual's genome.

NOTE: Having 0 pathogenic mutations does NOT completely eliminate the individual's risk of autism. Environmental factors, as well as other nonpathogenic genetic variants, also affect a person's risk of autism.

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RECOMMENDATIONS

• The CNTNAP2 gene encodes a protein that plays a part in the development of the nervous system. Your child's copy of this gene is statistically associated with a higher risk of autism.

- The CNTNAP2 gene tells your child's body to make a protein that is involved in the proper development of the nervous system. Specifically, it influences the way nerve cells organize themselves, conduct signals, and interact with supporting cells. Your child has a harmful change in this gene that's statistically associated with a higher risk of autism. However, carrying this harmful mutation alone doesn't mean your child has autism.
- In case you are interested in knowing about autism interventions for children Intervention strategies may vary by the age and strengths and weaknesses of the child. For example, intervention for a toddler with a recent diagnosis of ASD may include behavioral and developmental approaches (individually or in the context of comprehensive approach) and, as he progresses, involvement in a specialized or typical preschool program. For older children, intervention is more likely to occur in educational settings, with integration of behavioral and developmental therapies to promote skill development.
- Your child has a genetic mutation located near the ARID1B gene. This mutation is linked to mild mental impairment (MMI). However, MMI individuals can live independently and hold a job with some support from modern technology.
 - The ARID1B gene is linked to intelligence by way of affecting a person's corpus callosum. The corpus callosum is a structure in the brain that connects the left and right side of the brain, allowing for proper communication between the two halves. Albert Einstein, a genius physicist, had an unusually thick corpus callosum.
 - Your child carries a mutation located near the ARID1B gene that is linked to mild mental impairment. However, general research has shown that—over the past decades—the Asian population's average intelligence scores have increased and slightly surpassed that of many other groups. Therefore, this genetic result—by itself—doesn't mean your child carries a form of intellectual disability. The only way to be sure of your child's intelligence is to have him professionally evaluated by a team of experts, including a pediatrician or a psychologist, as the case requires.
 - As technology advances at a rapid pace, this may be helpful for people with mild mental impairment. Parent of the affected children should take the time to prepare their children with the skills they need to keep up with technological changes over time. To that end, parent often enroll their children in specialized programs that arm them with these skillsets, which may include an understanding of technology and how to persevere in the face of new challenges.
- Your child has a mutation in a brain development and maintenance gene that's associated with a higher risk of schizophrenia. Because of this, he may be more prone to having hallucinations.



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G-AUTISM REPORT | 21 August 2020

DOB: 10 October 1990

5/16

User: Nguyen Van A

Genetica ID: QWEKSU8UY6T **Gender**: M

- The CYFIP1 gene can be thought of as a brain cell development and maintenance gene. The protein encoded by this gene interacts with other proteins that might affect the way neurons (brain cells) grow appendages, called axons and dendrites, which are very important for the proper chemical communication between neurons. This same gene might also affect the upkeep of these appendages over time in the same manner. Scientists believe that a person's genes and differences in brain structure, function, or chemical communication might affect a person's chances of having schizophrenia.

- Schizophrenia is a serious and disabling mental disorder that alters how a person thinks, feels, behaves, and interacts with others. Affected children may have a difficult time differentiating reality from hallucinations. Additionally, they may experience delusions, have trouble thinking and focusing, and lack motivation. Within the East Asian population, specific symptoms may also include visions of Buddha and a belief that one is being haunted by an ancestor for their family's past wrongdoing.
- Our analysis shows that your child carries a mutation in his CYFIP1 gene that's associated with an increased risk of schizophrenia. Does this mean your child is certainly schizophrenic? The answer is no. In fact, studies suggest that this is a complex disorder that's involved by many other environment aspects, e.g. how a child is raised in a family.
- Children who have schizophrenia symptoms should see a doctor right away to get correctly diagnosed and treated. One type of treatment for this disorder is antipsychotic medications. These may be prescribed to help reduce symptoms, including hallucinations, paranoia, and unusual thinking. While side effects are common, stopping the medication abruptly can make these symptoms worse, so it's advised to strictly follow doctor's advice.
- Your child carries a genetic mutation in his genome that's linked to an increased risk of having autism spectrum disorder (ASD). Children with this disorder often face with social communication difficulties.
 - Our analysis detected a specific mutation in your child's DNA that may increase his risk of autism spectrum disorder (ASD). ASD refers to a group of complex developmental disorders that start in early childhood, and continues throughout a person's life. In some children, symptoms can improve as they get older.
 - Children with ASD often thrive on routine and tend to adhere to them very strictly. A deviation in their normal daily routine—such going out to eat at a restaurant instead of eating at home—can be extremely distressing for them. They may struggle to adapt to change, and might become frustrated or have "meltdowns".
 - For children who have social communication difficulties, applied Behavior Analysis can be an effective intervention. In general, this intervention is led by a trained therapist, who uses highly structured training sessions to help children build positive behaviors and reduce negative ones, like tantrums. The specific type of training will depend on the child's age and unique needs.
 - While your child carries an increased risk mutation of ASD, it doesn't mean that he is autistic. But it's important for you to monitor him, and contact his doctor, in case he develops atypical tendencies, especially if he has lot of difficulties in social communication.
- Your child carries a mutation in a brain development protein gene that is associated with speech delay that is inherent to autism. In particular, autistic children who carry this mutation also have higher chance of having language impairments.



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- The WNT2 gene encodes signaling proteins that may influence the development of the fetal brain (the development of a child's brain while he was still in the womb). Your child has a mutation in this gene that raises his risk of autism-related language impairment. So if your child had speech delay as an infant, there is a chance it is caused by this mutation.

- The Wnt signaling pathway also plays a role in the development of central nervous system. Previous evidence suggests that WNT2 regulates the development of midbrain and brainstem, which modulate auditory processing. Hence, the mutation in the WNT2 gene may perturb the neurodevelopmental process and interfere with auditory processing to hinder the language acquisition.
- If you suspect that your child has language-impairment issues, and a diagnosis of autism has not been excluded, it will be important for you to see a speech or language pathologist. Such a professional will be able to delineate whether your child's expressive and receptive language skills are appropriate. The pathologist will also be able to define for you if your child's use of pragmatic and conversational language is sufficient for his age.

GENES ANALYZED

48 genes associated with Autism Spectrum Disorder were analyzed.

AC005229.5, AC006974.1, AC006974.2, AC018450.1, AC019131.1, AC063965.2, AC073358.1, AC083849.1, AC110995.1, AC112656.1, AC245140.2, AL161747.1, AL590764.1, AP002387.1, AP003783.1, AUTS2, C7orf33, CHD8, CNTNAP2, CNTNAP2-AS1, DNASE1L1, DPY19L4P2, EI24P4, EIF4E, FLJ42102, GAPDHP47, KLLN, LOC101928700, MECP2, MED6P1, MIR718, NLGN3, NLGN4X, OPN1LW, PBX2P1, PTCHD1, PTCHD1-AS, PTEN, RAB2B, RNLS, RPL10, SHANK2, SHANK2-AS1, SLC9A9, SNORD8, TMLHE, TMLHE-AS1, WBSCR17

ELECTRONICALLY SIGNED BY

Duyen Bui, PhD, CSO & Lab Director on 21 August 2020 at 06:52:47

Rama Kota, PhD, Lead Geneticist on 21 August 2020 at 05:52:47

DOB: 10 October 1990

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What is autism?

Autism is more formally called autism spectrum disorder (ASD).

ASD refers to a range of previously distinct conditions that stem from issues with the development of the nervous system. 9,10 ASD affects approximately 1-2% of the world's population and is characterized by the following:10.11

- A. Persistent problems with two-way social communication and interactions
- B. Restricted and repetitive patterns of behavior or interests
- C. A and B are present from early childhood and hinder everyday life

Currently, autism has no known cure and cannot be diagnosed via a pregnancy-based test. 9.12

Autism is not mental retardation

Mental retardation has become an outdated term, and is now called intellectual disability.

Recent epidemiological studies suggest that roughly 30% of children with autism also have an intellectual disability. So, it's clear that autism and mental retardation can and do coexist—but importantly, they are not the same.

Autism spectrum disorders are developmental disabilities characterized by problems in communication, social interactions, and behavior. There is a wide "spectrum" of symptoms and varying levels of disability experienced among autistic individuals. Some show mild symptoms while others have severe ones.

Intellectual disability, on the other hand, often refers to below average intellectual abilities originating under the age of 18. It's characterized mainly by having an IQ score below 70, along with difficulties in life skills, such as self-care, communication, and interpersonal skills.

Therefore, a key distinction between autism and mental retardation is that people with autism do not always have diminished intelligence. In fact, some autistic individuals may have normal or very high IQs. However, it may be harder for us to know given their difficulties with communication and social skills—issues that are also present in mental retardation.

Asperger syndrome

Asperger syndrome is no longer a standalone condition. Instead, Asperger syndrome is now considered to be on the high functioning end of the autism spectrum.

Compared to others on the autism spectrum, people with Asperger syndrome have normal to higher intelligence and exceptional language skills. Although, many with Asperger syndrome tend to speak in a very particular or "robotic" manner. They may also not pick up on social cues, such as jokes or sarcasm, in group settings.

Perhaps the most prominent feature of Asperger syndrome is the person's obsession about a single object or subject. They'll want to learn as much as possible about it, and their thoughts and conversations often revolve around that particular thing of interest.

Other behaviors associated with Asperger syndrome include uncoordinated movements due to delayed motor development, such as having a "bouncy" walk, and repetitive routines.

While the cause of Asperger syndrome is still unknown, there seems to be a strong genetic basis.

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Williams syndrome

Williams syndrome is a developmental disorder caused by the deletion of genetic code from chromosome 7. This genetic condition is usually not inherited, and is characterized by unique facial features, mild to moderate intellectual deficits, and cardiovascular health issues.

People with Williams syndrome typically have outgoing, extremely friendly, and highly empathetic personalities. Given these prosocial behaviors, it would seem that Williams syndrome is quite the opposite of autism. However, researchers have discovered that individuals affected by Williams syndrome do, in fact, share some key similarities with those who fall on the autism spectrum: they both have problems with social interaction and social communication.

Overlapping symptoms between Williams syndrome and autism can include:

- Delayed language development
- Poor social skills
- Difficulty having conversations that involve thinking from different perspectives
- · Trouble with making and keeping relationships
- Obsessiveness, or limited interests
- Excessive fondness for routine
- · Inability to stay focused

How do genes affect autism?

To date, hundreds of genes have been linked to ASD via genetic studies on over 10,000 ASD patients and their families.13

While numerous factors contribute to the development of ASD, studies on siblings—especially twins—have shown that a person's genes play a large role. A recognized genetic basis for ASD is present in approximately 30% of people diagnosed with this condition.11

Recent research on Vietnamese children with ASD has confirmed the role of numerous—likely universal—genes implicated in ASD, including CHD8, DYRK1A, GRIN2B, SCN2A, OFD1 and MDB5. However, this same research has shown that the development of ASD in the Vietnamese population may be uniquely influenced by other genes, such as IGF1, LAS1L, and SYP.14

Mutations in these, and other genes, may negatively impact numerous aspects of the nervous system. For example, some of these mutations may alter the structure or function of synapses (connections) between neurons (brain cells), something implicated in the development of ASD.15

Genetica Autism Report description

This report decodes genetic variants/mutations that are associated with Autism Spectrum Disorders (ASD).

- Analyze 48 genes focusing on Pathogenic and Likely Pathogenic Mutations on ASD
- Analyze 1200+ variants to construct Polygenic Risk Score on ASD



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Genetica testing methodology

Saliva is collected using GeneFiX™ Saliva DNA Collector which can preserve the quality of the DNA in the collected saliva for at least 6 months at room temperature. The GeneFiX™ Saliva DNA Collector is developed, manufactured and monitored in accordance with the Quality Management System based on ISO 9001:2015 and ISO 13485:2016.

The genomic DNA is extracted from the collected saliva using Chemagic Prime™ Robot. The process is entirely automated using chemagen patented M-PVA Magnetic Bead technology for DNA and RNA purification with liquid handling to provide highthroughput automated isolation of ultra-pure nucleic acids. The process is monitored in accordance with the Quality Control of ISO/IEC 17025.

The extracted DNA is then enriched for targeted regions using a hybridization-based protocol and decoded using Genetica® V3 Proprietary Gene Decoding Chip. Genetica® V3 Gene Decoding Chip includes 800,000 single-nucleotide polymorphisms (SNPs), insertions or deletions (Indels) and copy-number variants (CNVs). Genetica uses its proprietary Artificial Intelligence Engine to extract the SNPs, Indels and CNVs from more than 435,000 published, scientific papers. The Artificial Intelligence Engine ranks the variants/mutations based on the significance and extracts the top 800,000 most significant variants/mutations for Asian

population. All pathogenic and likely pathogenic variants are characterized using Clinvar and ACMG databases. Genetica's workflow uses Thermo Fisher GeneTitan Platform and Illumina Hiseq 2000 (600 Gb in a single run). All samples are processed in the RUCDR Infinite Biologics Clinical Genomics Laboratory with CLIA-certified and CAP-accredited (CLIA Number: 31D2077913, CAP Number: 8981166).

Limitations

Variant of unknown significance (VUS) is a variation in a genetic sequence for which the association with disease risk is unclear. There is a possibility that a variant is characterized as VUS at the time this test may be characterized as benign or pathogenic in the future.

G-AUTISM REPORT

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21 August 2020

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DISCLAIMER

Gene Friend Way provides genetic assessment services for research or investigational use. Gene Friend Way does not provide any direct medical advice to individual patients. Genetic information must always be considered in conjunction with other information about your health such as lifestyle, family history, risk factors, biomedical data, diet, nutrition and physical activity among other factors.

Gene Friend Way's role is limited to providing results of genetic test and providing a broad set of general recommendations. More detailed recommendations that may be specific to you are to be made by qualified Professional Practitioners only. General guidelines provided in our report are for information purpose only and are meant to aid your Professional Practitioner to render the relevant professional or medical advice and treatment. While assessing your genetic parameters and providing the report and recommendations, we do not consider your past or existing health conditions and or any medication taken by you (either in the past or currently), even if you may have provided us with such information. Our report and the recommendations therein are to be acted upon in consultation with a medical or other health and wellness professional practitioner.

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PSYCHOLOGICAL DEVELOPMENT QUESTIONNAIRE FOR TODDLERS

MY CHILD...

1.	points or gestures to show interest or get attention.	Rarely	Sometimes	☐ Often
2.	has unusual or variable responses to sound (seems not to hear or is oversensitive or overreacts).	Rarely	Sometimes	☐ Often
3.	smiles or makes regular eye contact with others.	Rarely	Sometimes	Often
4.	responds to name when called.	Rarely	Sometimes	Often
5.	shows interest in children at play.	Rarely	Sometimes	Often
6.	enjoys doing "handshake" or "peek-a-boo."	Rarely	Sometimes	Often
7.	relates to others by babbling, gesturing, talking or changing expressions.	Rarely	Sometimes	☐ Often
8.	uses 3 or more words regularly and appropriately.	Rarely	Sometimes	Often
9.	speaks in phrases (for example: want juice, go bye-bye, more candy, give please).	Rarely	Sometimes	☐ Often
10.	laughs when others laugh.	Rarely	Sometimes	Often
11.	stopped using words or relating to others (at any time).		Sometimes	☐ Often

Source: Zahorodny, W., Shenouda, J., Mehta, U., Yee, E., Garcia, P., Rajan, M., & Goldfarb, M. Preliminary Evaluation of a Brief Autism Screener for Young Children. 2018. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5895140/



PARENTAL SURVEY

Check all that apply or attach recent clinical notes.

PARENT INFORMATION							
Name	DOB	Gender	Relationship with Proband				
Name	ров	F M	Mother Father				
Has autism? Yes No I don't	t know						
Has other developmental disorder Yes. S	Specify		No I don't know				
PROBAND INFORMATION							
Name	DOB	Gender ☐ F ☐ M	Ethnicity				
PROBAND CLINICAL HISTORY							
NEURODEVELOPMENT							
Developmental delay Yes No							
☐ Motor function ☐ Language learning	Global						
Intellectual disability Yes No Moderate	IQ Score Severe						
EPILEPSY							
Seizures Yes No Age at first unprovoked seizure:							
Seizures are Refractory Well-controlled							
NEURODEVELOPMENT	GRO	WTH					
☐ Hypotonia	Lo	w birth weight. Speci	fy				
☐ Hypertonia	☐ Fa	ilure to thrive					
Cerebral palsy	-	vergrowth					
☐ Encephalopathy	-	ort stature					
Structural brain anomaly							
Others	Head	circumierence					
HEARING / VISION Hearing loss. Specify Abnormal eye movement. Specify Abnomanility of vision. Specify Others	Pr Sp M	oling with autism enatal exposure to ai ecify aternal obesity, diabe	r pollution or certain pesticides. etes, or immune system ception				

Other services from Genetica®



G-Care

Foundation To Your Quality Life

Over 18 years of age

NutriCare

- Carbohydrate metabolism
- Protein metabolism
 Fat metabolism

HealthCare

- Breast Cancer or Prostate Cancer
- Stomach Cancer
 Asian Flush

Facts-to-Know

- Béo bụng
- Chuyển hóa affeineNguy cơ mất ngủ

- Belly fat
- Caffeine sensitivity
 Insomnia tendency

PhysiCare

- Endurance ability
- Power performance
 Cardiorespiratory fitness



G-Pro **Unearth Your Potentials**

Over 18 years of age

Mind & Spirit

- · Personality
- Behavior tendency
 Cognitive ability
- · IQ. EQ
- Educational attainment
 Language ability
- Math ability
- Music ability

Nutrition

- Protein, Fat, Carbohydrate
- metabolism

 Vitamin requirements
- · Liver Detoxification Food and Drink sensitivities
 Eating behavior
- Cardiometabolic risk
- Diabetes risk scores
 Cardiometabolic health

Fitness

- Endurance ability
- Power performance
 Cardiorespiratory fitness
- Recover ability
- Tendency to get injuries
 Weight Management Difficulty
 Tendon/ Ligament strength
- Exercise benefit

Resting

- · Insomnia tendency
- Belly fat
 Caffein metabolism

- Uterine, Kidney, Stomach, Leukemia,
 Liver, Lung, Pancreatic, Testicular,
 Prostate, Ovarian, Skin
 - Pheochromocytoma and
 Paraganglioma





G-Kid Care

Foundation For Child's Development

From 0 to 18 years of age

Behavioural tendencies

- Extraversion
- · Emotional instability

Health risk

· Obesity risk score

Macronutrient metabolism

- · Carbohydrate metabolism
- · Protein metabolism

Intelligence

- Cognitive ability





G-Kid Pro

Reaching An Optimal Future

From 0 to 18 years of age

Mind

- IQ
- · Educational attainment
- Cognitive ability
 Math ability
 Language ability
- Music ability
 Fitness potentials

Body

- Vitamin requirement
 Mineral requirement
- Macronutrients requirement

- Eating behavior
 Sweet and bitter taste
 Health risk: Obesity
 Cardiometabolic/ Diabetes risk.

- Personality
 Behavior tendency





Other services from Genetica®

gen<u>es</u>

G-Health

Access Risk Of Hereditary Diseases

• Ovarian

Pheochromocytoma
 and Paraganglioma

Over 18 years of age

Up to 20 common cancers for both genders

- Breast • Bladder
- Kidnev
- Brain
- Cervical

- Esophageal
- Colorectal
 Colon
- Uterine
- Stomach Leukemia Liver
- LungPancreatic
- Testicular Prostate





G-Immunity

Hereditary Susceptibility To **Respiratory Viral Infection Screening**

- · SARS-CoV · Influenza
- Acute Respiratory Distress Syndrome (ARDS)

Everyone will benefit from this report.

- Provide users the genetic information about their susceptibility to viral infection.
 Mainly focusing on respiratory viral infections at this time.
- Provide actionable guidelines and recommendations based on research studies from epidemics that have happened, such a SARS epidemic,





G-Stroke Hereditary Stroke Screening

The G-Stroke report will provide the following information:

• Pathogenic or likely pathogenic mutations that increase risk of strokes (ischemic stroke, hemorrhagic stroke).

• Stroke Risk Score: a polygenic score that indicates the patient's increased risk of

Everyone will benefit from this report.

- In-depth knowledge about gene-related causes of stroke.
- Information to make informed medical and lifestyle decisions in regard to the user's
- risk of stroke.

 Personalized recommendations for preventive and monitoring options.





G-ADHD

Hereditary ADHD Risk Screening

- The G-ADHD report will provide the following information:
 Pathogenic or likely pathogenic mutations that increase risk of ADHD.
- \bullet ADHD Risk Score: a polygenic score that indicates the user's increased risk of ADHD.

Young children and teenagers are especially advised to take the genetic test.

- In-depth knowledge about gene-related causes of ADHD.

 Information to help make informed medical and lifestyle decisions in regard to the user's risk of ADHD, as well as available treatment options.
- Helpful and personalized recommendations.

Thank you,

It is our honor to be able to contribute to your healthy and happy life.

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