

COMPREHENSIVE
canine DNA screen

ADVANCED GENOMICS TESTING FOR YOUR DOG'S HEALTH

REPORT FOR MORPHEUS





Canine DNA Screen

Report for

Morpheus

Basic Details

Breed:	Morpheus	Sex :	Male	Age :	2 weeks
Parents:	German Shepherd (M) German Shepherd (F)	Weight:	1 kgs	Height:	8 cm
Known health conditions:	No known health conditions specified		/		/

Sample Details

Reference Id:	REF21121001	Collected:	12/12/2021 5:30PM	Sample Type:	Swab Kit
Lab Ref Id:	SLCTD01001	Received:	13/12/2021 1:00PM	Reported:	26/12/2021

Reference Details

Referring Vet:	Dr. Shalini Upadhyay	Clinic's Name:	Animal Care Clinic
Clinic Address:	#14 Rajbhavan Road, Beside Yashoda Hospital,		
Owner name:	Kamakshi Setubandhu	Email Address:	k.setubandhu@gmail.com
Owner Address:	721, Block-D, Lake View Apartments, Rajbhavan Road, Somajiguda, Hyderabad, Telangana 500082		

Genomics Test Details

Panel:	Thermofisher Canine Genomics Traits and Disorders, Version 1 Panel - Canine Traits and Disorders, Version 1		
Laboratory:	GeneTech	Technology:	NGS
Machine:	Ion GeneStudio S5 and Ion Chef	LibPrep Solution:	Ampliseq based AgriSeq
Panel size:	154 markers 97 SNPs, 6 MNPs, Marker Coverage: 134 Genetic Disorders, 20 Traits 13 Ins, 38 Del		

SUMMARY OF RESULTS

Disorders 1	DISORDERS IDENTIFIED
	Achromatopsia-2
Carrier 2	CARRIER STATUS IDENTIFIED
	Amelogenesis Imperfecta Leukocyte Adhesion Deficiency, Type III

Traits	1
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TRAITS IDENTIFIED
Coat Color, Brown



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Introduction

Dogs are true companions of humans. There are many health issues that can affect dogs starting from common infections and allergies to complex genetic conditions which may set in early or could be late onset. It is vital to be aware of the health risks to the dog and provide care necessary for its healthier life and overall well-being.

About Canine DNA Screen

Canine DNA screen is a genomics test for dogs offered to pet owners and veterinarians. It allows identification of multiple diseases in a single test and makes it possible to profile the pets in advance, and provide better management and care for the pet. GeneTech's Canine DNA Screen genetic panel is based on ThermoFisher's Canine Disorders and Traits Panel which was developed based on extensive research in Canine Genomics and Veterinary Science.

The genomics panel offers screening for diseases recommended by ISAG (International Society for Animal Genetics) and more. The panel identifies selected 134 gene variants that are associated with common inherited conditions in different dog breeds along with 20 different variants associated with breed traits related to coat, hair and tail. Next generation sequencing (NGS) is used by the laboratory to identify gene variations or mutations.

Canine DNA Screen is a screening test and further confirmatory tests may be required based on clinical correlation and veterinarian's recommendations.

What is a Genetic Disorder?

A genetic disorder is a health condition caused by an abnormal genetic change usually called as mutation. Although some of these mutations occur spontaneously, a majority of them are inherited and are hereditary. Around 400 hereditary diseases known in canines are caused by mutations in 19,000 genes spread across 78 chromosomes. Genetic disorders either result in multi organ defects and metabolic dysfunction or may alter only one functional system of dog's health such as vision, skin, immune system, nerve and muscular function etc.

These genetic disorders are not uncommon and are not exclusive to pure breeds although they might be extensively reported in few selected breeds.

How is a genetic disorder inherited from a parent to the pup?

Inheritance is the process by which genetic information is passed on from parent to offspring. Nature provides two copies of gene for all bodily functions of which one copy comes from father and other from mother. Every genetic disorder inherits in a specific manner as given below:

- **Autosomal Dominant:** If a disease is expressed in a dog with one copy of abnormal gene and one copy of normal gene. The risk on passing on such a disease to offspring is 50%.
- **Autosomal Recessive:** If a disease is expressed in a dog only if both copies of the gene are abnormal. Such a pattern is common with 25% risk to offspring if both parents are unaffected carriers with one abnormal gene copy each.
- **X-linked:** The disease is expressed only in male dogs and female dogs are unaffected carriers.
- **Complex:** The inheritance of disease is called complex when it results due to multiple genes and their interaction with environmental factors.

There are some disorders for which the inheritance pattern is unknown and a few with more than one inheritance pattern associated.

How does Canine DNA Screen help?

- Early diagnosis and preventive care are possible before the symptoms set in
- Potential dog owners may choose to adopt a dog based on test result
- Carrier status for diseases can be identified, which allows choice of right parents in breeding programs to reduce disease risk in pups
- Veterinarians can offer evidence based, effective management and treatment to the dog



A GUIDE TO THE DNA SCREENING REPORT



Introduction to genetic terms used in the report

DNA is the genetic material present in every cell containing a code that controls various functions in your pet's body.

Gene is a unit of DNA with genetic information or code to produce a specific protein or an enzyme in your pet's body to perform a specific function.

Gene Mutations or Gene Variants are specific changes of the genetic code at specific locations on the genes. These change of code result in a protein variation resulting in a health issue.

Genetic Disease or Disorder is a particular abnormal health condition affecting function of a body or a part of dog's body due to a genetic change. Based on the organ system involved, dog's genetic disorders can be one the following types:

How does Canine DNA Screen help?

- Ophthalmic (Vision problems related to eye)
- Dental (Teeth and denture related)
- Neurological (Disorders of the nervous system)
- Immunological (Conditions with immune dysfunction)
- Endocrine (hormonal or endocrine disorders)
- Skeletal (Bone defects)
- Respiratory (Breathing conditions)
- Hepatological (Liver disorders)
- Dermatological (Skin conditions)
- Metabolic (Abnormal body metabolism or enzyme defects)
- Hematological (Blood tissue related disorders)
- Gastrointestinal (Digestive system defects)
- Muscular (Muscle dysfunction)
- Renal (Kidney and urinary defects)

Genetic Trait is a distinguishing quality or characteristic of the dog determined by genetic factor or change.

Autosomal: Dogs have 78 chromosomes (39 pairs) with 38 pairs are autosomes (non-sex chromosomes) and one pair of sex chromosomes (X and Y).

Inheritance is the process by which genetic information is passed on from parent to offspring. Nature provide two copies of gene for all bodily functions of which one copy comes from father and other from mother.

How to read Canine DNA Screen Result?


We have designed this report carefully to provide you with as much information as possible in a simple yet comprehensive fashion. The detailed Table of Contents page guides you to various parts of the report.




Canine DNA Screen aims at identification of the following by testing 154 genetic markers:

- Is your dog affected with any of the inherited genetic conditions tested?
- If your dog is an unaffected carrier for any of these conditions?
- What are the genetic traits (among those the panel tests) that your dog has inherited?

[illegible][illegible][illegible]



University of Lincoln
ACADEMIC YEAR 2023-2024



UNIVERSITY OF LINCOLN

STUDENT INFORMATION

Name	Matriculation Number	Date of Birth	Gender

DEGREE COURSE

Degree Course	Semester

ABOUT THE COURSE

This programme is an online degree programme that allows students to study for a Bachelor's degree in Information Management. The programme is designed to provide students with the knowledge and skills required to manage information systems and to develop their critical thinking and problem-solving abilities. The programme is delivered through a combination of online lectures, self-paced modules, and practical exercises. Students are required to complete a final project and a dissertation as part of the programme. The programme is accredited by the Quality Assurance Agency for Higher Education (QAA) and is recognised by the Higher Education Funding Council for England (HEFCE).

COURSE AND PROGRAMME

The programme is a three-year programme that is delivered through a combination of online lectures, self-paced modules, and practical exercises. The programme is designed to provide students with the knowledge and skills required to manage information systems and to develop their critical thinking and problem-solving abilities. The programme is delivered through a combination of online lectures, self-paced modules, and practical exercises. Students are required to complete a final project and a dissertation as part of the programme. The programme is accredited by the Quality Assurance Agency for Higher Education (QAA) and is recognised by the Higher Education Funding Council for England (HEFCE).

DEGREE DEDUCTION

The programme is a three-year programme that is delivered through a combination of online lectures, self-paced modules, and practical exercises. The programme is designed to provide students with the knowledge and skills required to manage information systems and to develop their critical thinking and problem-solving abilities. The programme is delivered through a combination of online lectures, self-paced modules, and practical exercises. Students are required to complete a final project and a dissertation as part of the programme. The programme is accredited by the Quality Assurance Agency for Higher Education (QAA) and is recognised by the Higher Education Funding Council for England (HEFCE).

QUALIFICATION AND AWARD

The programme is a three-year programme that is delivered through a combination of online lectures, self-paced modules, and practical exercises. The programme is designed to provide students with the knowledge and skills required to manage information systems and to develop their critical thinking and problem-solving abilities. The programme is delivered through a combination of online lectures, self-paced modules, and practical exercises. Students are required to complete a final project and a dissertation as part of the programme. The programme is accredited by the Quality Assurance Agency for Higher Education (QAA) and is recognised by the Higher Education Funding Council for England (HEFCE).

REQUIREMENTS

1. A minimum of 120 credits at the end of the first year of the programme.
2. A minimum of 120 credits at the end of the second year of the programme.
3. A minimum of 120 credits at the end of the third year of the programme.
4. A minimum of 120 credits at the end of the fourth year of the programme.

REFERENCES

1. University of Lincoln, *Information Management*, (Lincoln: University of Lincoln, 2023).
2. University of Lincoln, *Information Management*, (Lincoln: University of Lincoln, 2023).
3. University of Lincoln, *Information Management*, (Lincoln: University of Lincoln, 2023).
4. University of Lincoln, *Information Management*, (Lincoln: University of Lincoln, 2023).

Results Summary Page

Detailed Results Page

List of Traits Tested

List of Disorders Tested



**Ophthalmic
ACHROPSIDIOSIS-2**



DISEASE CONDITION

Mnemonic	ACIDGTONS	Gene Name	Cyclic Nucleotide Gated Channel Alpha 3	Gene	CNGA3
Category	Congenital	OMIM	266500	OMIM	266500
Cure	CMSD	Genotype	Reference Q	Variant Found A	CNGA3 c.1042G>A

About the Condition

Achroplasia, also called as day blindness or color blindness, is characterized by complete or partial loss of cone vision, leading to severely reduced or complete vision loss during the light hours. Other signs/symptoms of evening bright light or glare in bright lights (photophobia), nyctalopia (supernormal eye movement) and sometimes total color blindness. CNGA3 gene associated achroplasia is a rare genetic condition with complete loss of cone photoreceptor function. The cone photoreceptors transmit signals. Current development may also occur regularly in affected dogs.

Cause and Prognosis

Symptoms appear early between 0-2 weeks of age when owner observes gradually difficulty in registering achromats. Initially cones start developing but since from the onset, cone and rod segments gradually deteriorate, followed by a slow loss of cones throughout the dog's lifetime. Cone function loss is confirmed by electroretinography.

Dog Breeds

CNGA3 gene associated congenital achroplasia is reported in German Shepherds, Labrador Retrievers, Golden Retrievers.

Genetics and Inheritance

Mutation in CNGA3 gene causes dysfunction of retinal phototransduction pathway, leading to congenital achroplasia. It is an autosomal recessive disorder, resulting when both copies of CNGA3 gene are mutated, one copy inherited from each parent. Unaffected carrier parents have 25% risk of having a pup with achroplasia. 2.

Management

- Although currently, there is no cure, clinical trials in the area of gene therapy are showing encouraging results for treating CNGA3 associated achroplasia. 2
- Animal additional changes to assist by wearing the dog in shady regions, evening brights to direct sunlight, regular optometric checkups and electroretinography to monitor by Veterinarian.
- Avoid using affected dogs and unaffected carrier parents in breeding programs.

References

1. Taylor, M.S., D'Silva, E.M., D'Amico, S., D'Amico, S., MacDonald, A.C., Saunders, S.L., Chinnai, M.R., Chinnai, C.J., Cavell, M.L., Allen, M.L., Aguirre, G.D., Venkatesh, S.G., Gnanapavan, K.L. **Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achroplasia Associated Cyclic Nucleotide Gated Channel Alpha 3 Deficiency**. *PLoS One* 10(10):e0140883, 2015.

DISORDER DETAILS PAGE

One page for each positive marker identified. Find details below on how to read/use this page.

Marker details table

Marker details table
Provides details about the marker that tested positive along with full gene name.

Onset and Prognosis

Onset and Prognosis
Not all genetic diseases express at birth and presentation of symptoms may vary within the breed or across breeds. Progression of the disease may be slow or rapid. This section describes what is known about this condition in this regard.

Dog Breeds

Dog Breeds
Some diseases are widely reported in certain breeds which are mentioned in this section.

Management

Management

Disease management is provided in a distilled fashion in this section. Preventive measures, dietary management, medical care, and promising treatments in development are covered.

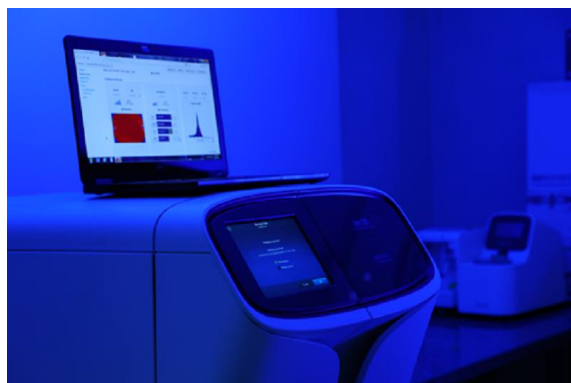
Discuss your pet's report with veterinarian before implementing any of the recommendations provided in this section.



Limitations of Canine DNA Screen report

Product and Technology Limitations

- The test is limited to the genes and diseases tested in the panel. Genetic disorders associated with genes not included in the panel cannot be diagnosed.
- A genetic disease can be caused by multiple variants of a gene. However, only selected variants (which are commonly reported for certain breeds) are included in the panel for testing. In case your pet is clinically showing symptoms of any genetic condition and your Canine DNA Screen report is negative for the disease, please consult our geneticist for available confirmatory options.
- The panel is based on current research. More and more genes and their association with diseases are being discovered. Newer versions of the panel will be released periodically and shall be used. Write to us if you need more information on this.
- Complex polygenic genetic disorders caused by multiple genes are not covered in this test.
- Clinical correlation of the genetic report is strongly recommended.
- Recommendations provided in the report are general. Veterinarian consultation is mandatory.
- Next Generation Sequencing technology was used to generate the data for analysis. Next Generation Sequencing (NGS) is the latest technology for multi-gene sequencing with 99% accuracy and sensitivity.





Results detailed report

Summary of results identified

Disorders 1	Disorders identified Achromatopsia-2 Type: Ophthalmic Marker: AGSCTD079 Gene: CNGA3 Inheritance: Autosomal Recessive Summary: Achromatopsia 2 is an inherited eye disease, characterized by early onset loss of cone photo-receptor function resulting in day blindness, total color blindness, decreased central visual acuity.
Carrier 2	CARRIER STATUS IDENTIFIED Amelogenesis Imperfecta Type: Dental Marker: AGSCTD003 Gene: ENAM Inheritance: Autosomal Recessive Summary: Amelogenesis Imperfecta is a common non-syndromic genetic condition caused by malfunction of enamel proteins, resulting in enamel hypoplasia or thinning, small and pointed teeth, rough surface with brown color, and greater gaps between teeth. Leukocyte Adhesion Deficiency, Type III Type: Immunological Marker: AGSCTD026 Gene: FERMT3 Inheritance: Autosomal Recessive Summary: Canine leukocyte adhesion deficiency is a primary immunodeficiency disorder characterized by recurrent bacterial infections in the presence of marked leukocytosis, impaired wound healing, fever, gingivitis, lameness, and enlarged lymph

nodes.

Traits

1

TRAITS IDENTIFIED

Coat Color, Brown

Marker: AGSCTD002

Gene: TYRP1

Inheritance: Autosomal Recessive

Summary: Coat color, brown is an inherited trait influenced by the pigment eumelanin which causes the darkening of a dog's coat color. This brown color coat is also referred to as liver, red or chocolate coat.

Details for each disorder or trait are provided in following pages.



Ophthalmic Achromatopsia-2

Marker:	AGSCTD079	Gene Name:	Cyclic Nucleotide Gated Channel Alpha 3	Gene:	CNGA3
Category:	Ophthalmic	Condition:	Achromatopsia-2	Inheritance:	Autosomal Recessive
Chr#:	CHR10	Genotype	Reference:	Var Found:	OMIA #:

About the Condition

Achromatopsia, also called as day blindness or color blindness, is characterized by cone photoreceptor dysfunction of the eye, leading to severely reduced or complete vision loss during day light hours. Dogs show signs of avoiding bright light or distress in bright light (photophobia), nystagmus (uncontrolled eye movements) and sometimes total colour-blindness. CNGB3 gene associated Achromatopsia is a rare genetic condition with complete loss of cone photoreceptor function while rod photoreceptors remain intact. Cataract development may also occur frequently in affected dogs.

Onset and Prognosis

Symptoms appear early between 8-12 weeks of age when owner observes pup's difficulty in negotiating obstacles. Initially cones start developing but once non-functional, their inner and outer segments gradually deteriorate, followed by a slow loss of cones throughout the dog's lifetime. Cone function loss is confirmed by electroretinography.

Dog Breeds

CNGA3 gene associated congenital Achromatopsia is reported in German Shepherds and Labrador Retrievers.

Genetics and Inheritance

Mutation in CNGA3 gene causes dysfunction of retinal phototransduction pathway, leading to congenital Achromatopsia-2. It is an autosomal recessive disorder, resulting when two copies of CNGA3 gene carry the mutation, one copy inherited from each parent. Unaffected carrier parents have 25% risk of having a pup with Achromatopsia-2.

Management

1. Although currently, there is no cure, clinical trials in the area of gene therapy are showing encouraging results for treating CNGA3 associated Achromatopsia-2. 2. Avoid additional damage to retina by walking the dog in shady regions, avoiding bright and direct sunlight. 3. Regular ophthalmic check-ups, electroretinography as directed by Veterinarian 4. Avoid using affected dogs and unaffected carrier parents in breeding programs

References

1.Tanaka, N., Dutrow, E.V., Miyadera, K., Delemotte, L., MacDermaid, C.M., Reinstein, S.L., Crumley, W.R., Dixon, C.J., Casal, M.L., Klein, M.L., Aguirre, G.D., Tanaka, J.C., Guziewicz, K.E.: **Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment.**PLoS One 10:e0138943, 2015.