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COMPREHENSIVE canine DNA screen

ADVANCED GENOMICS TESTING FOR YOUR DOG'S HEALTH

REPORT FOR SANJI

POWERED BY GeneTech

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Canine Traits and Disorders

Report for

Sanji

Basic Details

Breed:	**Labrador**	Sex :		Age :	
Parents:		Weight:		Height:	
Known Health Conditions :		**None**			

Sample Details

Reference Id:	**IonCode_0791**	Collected:	**20/11/2022**	Sample Type:	**Swab**
Lab Ref Id:	**BSCTD22K7**	Received:	**22/11/2022**	Reported:	**20/12/2022**

Reference Details

Referring Vet:		Clinic's Name:	
Clinic Address:			
Owner name:		Email Address:	
Owner Address:			

Genomics Test Details

Panel:	**Thermofisher Canine Genomics 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, 13 Ins, 38 Del**	Marker Coverage:	**134 Genetic Disorders, 20 Traits**

SUMMARY OF RESULTS

Disorders	**1**
Carriers	**3**
Traits	**4**

DISORDERS IDENTIFIED
Menkes Disease

CARRIER STATUS IDENTIFIED
Achromatopsia-2
Cone Rod Dystrophy 3
Osteogenesis Imperfecta SERPINH1 related

TRAITS IDENTIFIED
Coat colour, dominant black
Coat color grizzle, coat color extension
Coat color Agouti
Coat color extension

Neurological
Menkes Disease

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DISEASE CONDITION

Marker	**AGSCTD030**	GeneName	**ATPase, Cu transporting a-polypeptide**		Gene	**ATP7A**
Category	**Neurological**	Condition	**Menkes Disease**		Inheritance	**X-linked recessive**
Chr#:	**CHR X**	Genotype	Reference: **C**	Var Found: **T**	OMIA #	**000640**

About the Condition

Menkes disease, involving ATP7A, is a neurodegenerative disorder of copper deficiency. It is characterized by cerebral and cerebellar degeneration, connective tissue abnormalities, coarse hair and a failure to thrive. Symptoms are directly related to the dysfunction of copper dependent enzymes and there is a high variation in the severity of symptoms and if left untreated, may result in severe neurologic defects. The diagnosis is usually confirmed by analysing copper level in the urine.

Onset and Prognosis

This occurs at an early age and can be lethal if it is not treated. Phenotypic variability is reported.

Dog Breeds

Menkes disease is seen in Labrador Retriever, Doberman Pinscher, American Cocker Spaniel, Keeshond, Skye Terrier and West Highland White Terrier

Genetics and Inheritance

ATP7A gene is responsible for production of the ATPase enzyme that regulates copper levels in the body. The missense mutation causes decrease in copper levels and results to Menkes disease. The condition is inherited in an X-linked recessive pattern and affects only male dogs. Female dogs are unaffected carriers.

Management

1. Frequent copper analysis followed by early treatment can alleviate the neurological symptoms.
2. Avoid using affected dogs and unaffected carrier females in breeding programs

References

1.Wu, X., den Boer, E.R., Vos-Loohuis, M., Steenbeek, F.G.V., Monroe, G.R., Nijman, I.J., Leegwater, P.A.J., Fieten, H.**: Investigation of genetic modifiers of copper toxicosis in Labrador Retrievers**Life (Basel) 10:, 2020.

2.Pindar, S., Ramirez, C**: Predicting copper toxicosis: relationship between the ATP7A and ATP7B gene mutations and hepatic copper quantification in dogs**Hum Genet :, 2019.

3.Fieten, H., Gill, Y., Martin, A.J., Concilli, M., Dirksen, K., van Steenbeek, F.G., Spee, B., van den Ingh, T.S., Martens, E.C., Festa, P., Chesi, G., van de Sluis, B., Houwen, R.H., Watson, A.L., Aulchenko, Y.S., Hodgkinson, V.L., Zhu, S., Petris, M.J., Polishchuk, R.S., Leegwater, P.A., Rothuizen, J.**: The Menkes and Wilson disease genes counteract in copper toxicosis in Labrador retrievers: a new canine model for copper-metabolism disorders**Dis Model Mech 9:25-38, 2016.

Ophthalmic
Achromatopsia-2

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UNAFFECTED CARRIER

Marker	**AGSCTD079**	GeneName	**Cyclic Nucleotide Gated Channel Alpha 3**		Gene	**CNGA3**
Category	**Ophthalmic**	Condition	**Achromatopsia-2**		Inheritance	**Autosomal Recessive**
Chr#:	**CHR10**	Genotype	Reference: **G**	VarFound: **A**	OMIA #	**001481**

Please note that the dog is an unaffected carrier for the condition. Avoiding carrier in breeding programs is advised.

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), nystagamus (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. Diagnosis is by ophthalmic evaluation and electroretinography.

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 GermanShepards 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.

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.

Opthalmic
Cone Rod Dystrophy 3

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UNAFFECTED CARRIER

Marker	**AGSCTD051**	GeneName	**ADAM Metallopeptidase Domain 9**		Gene	**ADAM9**
Category	**Opthalmic**	Condition	**Cone Rod Dystrophy 3**		Inheritance	**Autosomal Recessive**
Chr#:	**CHR16**	Genotype	Reference: **C**	VarFound: **T**	OMIA #	**1520**

Please note that the dog is an unaffected carrier for the condition. Avoiding carrier in breeding programs is advised.

About the Condition

Code Rod Dystrophy-3, also called progressive retinal atrophy is an inherited genetic disorder characterized by blindness due to degeneration of photoreceptors, cones and rods. The condition starts with degeneration of cone first followed by the rods eventually resulting in complete vision loss. Symptoms of affected dogs include loss of peripheral and general vision, loss of color vision and photophobia. The disease can be diagnosed by opthalmoscopy and electroretinography.

Onset and Prognosis

The age of onset of the condition can be between 7 months to 1 year beginning with peripheral vision loss followed by complete vision loss over several years.

Dog Breeds

Code Rod Dystrophy-3 is seen reported in Glen of Imaal Terrier

Genetics and Inheritance

References

1.Goldstein O, Mezey JG, Boyko AR, Gao C, Wang W, Bustamante CD, Anguish LJ, Jordan JA, Pearce-Kelling SE, Aguirre GD, Acland GM.**: An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9. Mol Vis**2010 Aug 11;16:1549-69.

2.Kropatsch, R., Petrasch-Parwez, E., Seelow, D., Schlichting, A., Gerding, WM., Akkad, DA., Epplen, JT., Dekomien, G**: Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene**Mol Cell Probes 24:357-63, 2010.

Orthopedic
Osteogenesis Imperfecta SERPINH1 related

UNAFFECTED CARRIER

Marker	**AGSCTD116**	GeneName	**Serpin peptidase inhibitor, clade H (heat shock protein 47) member1, (collagen binding protein1)**		Gene	**SERPINH1**
Category	**Orthopedic**	Condition	**Osteogenesis Imperfecta SERPINH1 related**		Inheritance	**Autosomal Recessive**
Chr#:	**CHR21**	Genotype	Reference: **A**	VarFound: **G**	OMIA #	**001483**

Please note that the dog is an unaffected carrier for the condition. Avoiding carrier in breeding programs is advised.

About the Condition

Osteogenesis Imperfecta, SERPINH1 related is an inherited skeletal disease characterized by fragile bones. The condition, which presents from birth is also known as Brittle bone disease and is caused due to poor collagen formation. The symptoms include spontaneous fracturing of the bones and teeth, loose joints, difficulty walking, pain, osteopenia, blue eye sclera, stunted growth, weak tendons and muscle atrophy. In severe extreme cases ribs can fracture leading to difficulty in breathing. The disease can be diagnosed through X-rays and radiography.

Onset and Prognosis

This disease has an early age of onset and the affected dogs usually do not survive beyond a few weeks.

Dog Breeds

The condition is reported in Dachshund.

Genetics and Inheritance

The SERPINH1 gene encodes the protein Collagen which is crucial in bone formation. A frame shift mutation on the gene leads to Osteogenesis Imperfecta by disrupting collagen folding. It is an autosomal recessive disorder, resulting when two copies of SERPINH1 gene carry the mutation, one copy inherited from each parent. Unaffected carrier parents have 25% risk of having a pup with Osteogenesis Imperfecta SERPINH1 related.

References

1.Drogemuller, C., Becker, D., Brunner, A., Haase, B., Kircher, P., Seeliger, F., Fehr, M., Baumann, U., Lindblad-Toh, K., Leeb, T.**: A missense mutation in the SERPINH1 gene in Dachshunds with osteogenesis imperfecta**PLoS Genet 5:e1000579, 2009.

2.Eckardt, J., Kluth, S., Dierks, C., Philipp, U., Distl, O.**: Population screening for the mutation associated with osteogenesis imperfecta in dachshunds**Vet Rec 172:364, 2013.

3.Lindert, U., Weis, M.A., Rai, J., Seeliger, F., Hausser, I., Leeb, T., Eyre, D., Rohrbach, M., Giunta, C.**: Molecular consequences of the SERPINH1/HSP47 mutation in the Dachshund natural model of osteogenesis imperfecta**J Biol Chem 290:17679-89, 2015.

Trait
Coat colour, dominant black

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TRAIT

Marker	**AGSCTD044**	GeneName	**Beta-defensin 103**		Gene	**CBD103**
Category	**Trait**	Condition	**Coat colour, dominant black**		Inheritance	**Autosomal Dominant**
Chr#:	**CHR16**	Genotype	Reference: **CCC**	Var Found: **--**	OMIA #	**001416**

About the Trait

Coat color black is an inherited trait. The dominant black gene is due to a mutation in a Beta-defensin gene (CBD103). Coat coloration is controlled by several different genes in dogs. . The pigment Eumelanin can modify and create other colors such as liver (brown), blue (grey), or isabella (pale brown). Note that coat color is a complex multigene trait.

Dog Breeds

Coat color black is reported in almost all dog breeds.

Genetics and Inheritance

The gene CBD103 plays a role in pigmentation of cells and a 3 base pair deletion results in black coat. The trait follows autosomal dominant pattern with only one allele with mutation enough to express the trait.

References

1.Brancalion, L., Haase, B., Wade, C.M.**: Canine coat pigmentation genetics: a review**Anim Genet :, 2021.

2.Candille, SI., Kaelin, CB., Cattanach, BM., Yu, B., Thompson, DA., Nix, MA., Kerns, JA., Schmutz, SM., Millhauser, GL., Barsh, GS. A {beta}-Defensin Mutation Causes Black Coat Color in Domestic Dogs. Science 318**: 1418-23, 2007. Pubmed reference: 17947548 DOI:10.1126/science**1147880

Trait
Coat color grizzle, coat color extension

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TRAIT

Marker	**AGSCTD014**	GeneName	**Melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)**		Gene	**MC1R**
Category	**Trait**	Condition	**Coat color grizzle, coat color extension**		Inheritance	**Autosomal**
Chr#:	**CHR 5**	Genotype	Reference: **G**	Var Found: **A**	OMIA #	**001199, 001495**

About the Trait

Grizzle is a color pattern that appears as mixed hair on the dog with no discernible pattern. The color appears blended together, making it look like just one color until you examine it closely. Grizzle coloring may mix black hair with some tan or brown hairs, or white with black, making it appear grey. Coat color is a complex multigene trait.

Dog Breeds

The trait is reported in Irish Setter, Labrador Retriever, Australian Cattle Dog, Alaskan Husky, Siberian Husky and Afghan Hound

Genetics and Inheritance

The gene MC1R gene provides instructions for melanocortin 1 receptor which plays a significant role in pigmentation of cells. A missense mutation on the gene may result in Grizzle color coat.

References

1.Brancalion, L., Haase, B., Wade, C.M.**: Canine coat pigmentation genetics: a review**Anim Genet :, 2021.

2.Dürig, N., Letko, A., Lepori, V., Hadji Rasouliha, S., Loechel, R., Kehl, A., Hytönen, M.K., Lohi, H., Mauri, N., Dietrich, J., Wiedmer, M., Drögemüller, M., Jagannathan, V., Schmutz, S.M., Leeb, T.**: Two MC1R loss-of-function alleles in cream-coloured Australian Cattle Dogs and white Huskies**Anim Genet 49:284-290, 2018.

3.Dreger, DL., Schmutz, SM.**: A new mutation in MC1R explains a coat color phenotype in 2 "Old" breeds: Saluki and Afghan Hound**J Hered :, 2010.

Trait
Coat color Agouti

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TRAIT

Marker	**AGSCTD011**	GeneName	**Aspartic Peptidase, reteroviral like 1**		Gene	**ASIP**
Category	**Trait**	Condition	**Coat color Agouti**		Inheritance	**Autosomal**
Chr#:	**CHR 24**	Genotype	Reference: **TCTCA**	Var Found: **GCTCG**	OMIA #	**000201**

About the Trait

Coat color, Agouti is an inherited fur coloration displaying two or more bands of pigmentation. As a result the overall appearance of agouti fur is usually gray or dull brown and sometimes dull yellow. Agouti protein controls the release of melanin pigments (Eumelanin and Phaeomelanin) into the hair. A multi nucleotide variation leads to presence of display of two or more bands of pigmentation. Coat color is a complex multigene trait.

Dog Breeds

This trait is reported in German shepherd.

Genetics and Inheritance

The gene ASIP (Aspartic Peptidase, retroviral like 1) codes for Agouti-signaling protein which is responsible for the distribution of melanin pigment resulting in display of two or more bands of pigmentation. Inheritance pattern is complex, polygenic and autosomal in nature.

References

1.Dreger, D.L., Schmutz, S.M.**: A SINE insertion causes the black-and-tan and saddle tan phenotypes in domestic dogs**J Hered :S11-8, 2011.

2.Dreger, D.L., Parker, H.G., Ostrander, E.A., Schmutz, S.M.**: Identification of a mutation that is associated with the saddle tan and black-and-tan phenotypes in Basset Hounds and Pembroke Welsh Corgis**J Hered 104:399-406, 2013.

Trait
Coat color extension

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TRAIT

Marker	**AGSCTD013**	GeneName	**Melanocortin 1 receptor (alpha melanocyte stimulating hormone receptor)**		Gene	**MC1R**
Category	**Trait**	Condition	**Coat color extension**		Inheritance	**Autosomal Recessive**
Chr#:	**CHR 5**	Genotype	Reference: **C**	Var Found: **T**	OMIA #	**001199**

About the Trait

Coat color extension is an inherited trait. The E (extension) locus is governed by the MC1R gene creates the black facial mask of many dogs as well as yellow or red coats. MC1R activation prompts the melanocyte to produce eumelanin, whereas MC1R inhibition leads to the production of pheomelanin. A mutation of the MC1R gene can cause a dog's cells to only produce phaeomelanin in place of eumelanin. Mutations in MC1R have been associated with white coloring or partial red coat in several species. Coat color is a complex multigene trait.

Dog Breeds

The trait is reported in Irish Setter, Labrador Retriever, Australian Cattle Dog, Alaskan Husky and Siberian Husky

Genetics and Inheritance

The gene MC1R gene provides instructions for making a protein called the melanocortin 1 receptor. A non sense mutation inhibits protein Melanocortin making the coat color white or dull. Inheritance is not clear with both Autosomal dominant and recessive patterns reported.

References

1.Dürig, N., Letko, A., Lepori, V., Hadji Rasouliha, S., Loechel, R., Kehl, A., Hytönen, M.K., Lohi, H., Mauri, N., Dietrich, J., Wiedmer, M., Drögemüller, M., Jagannathan, V., Schmutz, S.M., Leeb, T.**: Two MC1R loss-of-function alleles in cream-coloured Australian Cattle Dogs and white Huskies**Anim Genet 49:284-290, 2018.

2.Nowacka-Woszuk, J., Salamon, S., Gorna, A., Switonski, M.**: Missense polymorphisms in the MC1R gene of the dog, red fox, arctic fox and Chinese raccoon dog**J Anim Breed Genet 130:136-41, 2013.