

Optimal Selection™

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LABORATORIES

BR00 274

Perfect Octave, Mastiff

Registered Name: Perfect Octave

Owner: Joseph Moody

Nickname: Octave

Country: United States

Registration ID: WS44065807

Testing date: 2016/8/19

Breed: Mastiff

Gender: Male

Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Canine Multifocal Retinopathy 1, (CMR1); mutation originally found in Mastiff-related breeds	Ocular Disorders	Autosomal Recessive	Carrier
Degenerative Myelopathy, (DM; SOD1A)	Neurological Disorders	Autosomal Recessive (Incomplete Penetrance)	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Ocular Disorders	Autosomal Dominant	Clear

Test results for pharmacogenetics

Disorder	Mode of Inheritance	Result
Multi-Drug Resistance 1, (MDR1)	Autosomal Dominant	Clear

On behalf of Genoscoper Laboratories,


SIGNATURE

Jonas Donner, PhD, Head of Research and Development
at Genoscoper Laboratories

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Test results - Traits - page 1

Coat Type

Trait	Genotype	Description
Coat Length	L/L	The dog is likely to have short-haired coat.
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/TC	The dog is not genetically likely to express furnishings.
<i>KRT71</i> c.451C>T (p.Arg151Trp)	C/C	The dog does not carry any copies of the tested allele causing curly coat. The dog most likely has non-curly hair.

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Test results - Traits - page 2

Coat Colour

Trait	Genotype	Description
Colour Locus E - Extensions	Em/Em	The dog is likely to have a dark mask.
Colour Locus B - Brown	B/B B/bd bd/bd	The dog doesn't have any of the tested b alleles causing brown pigment.
Colour Locus K - Dominant Black	ky/ky	The dog is likely to express the coat colour defined by the colour locus A.
Colour Locus A - Agouti	ay/ay	The dog is genetically sable.
Colour Locus S - Piebald or extreme white spotting	S/sp	The dog is likely to have solid coat colour or few white spots in its coat.
Colour Locus H - Harlequin	h/h	The dog doesn't have harlequin pattern.
Merle (M allele)	m/m	The dog is genetically non-merle and does not carry a <i>SILV</i> gene SINE insertion.
Saddle Tan (<i>RALY</i> gene dupl.)	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
Albinism (caL-allele)	C/C	The dog does not carry the tested mutation for albinism.

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Test results - Traits - page 3

Body Size

Trait	Genotype	Description
<i>IGF1</i> (chr15:41221438)	G/G	The dog is homozygous for the ancestral allele typically associated with large body mass.
<i>IGF1R</i> c.611G>A (p.Arg204His)	G/G	The dog carries two ancestral alleles typically found in larger-sized breeds.
<i>FGF4</i> insertion	D/D	The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length.
<i>STC2</i> (chr4:39182836)	T/T	The dog has two copies of the ancestral allele associated with larger body size.
<i>GHR1</i> (p.Glu191Lys)	G/G	The dog has two copies of the ancestral allele associated with larger body size.
<i>GHR2</i> (p.Pro177Leu)	C/C	The dog has two copies of the ancestral allele associated with larger body size.
<i>HMGA2</i> (chr10:8348804)	G/G	The dog has two copies of the ancestral allele associated with larger body size.

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Test results - Traits - page 4

Morphology

Trait	Genotype	Description
<i>BMP3</i> c.1344C>A (p.Phe448Leu)	C/C	The dog does not carry the tested allele typically associated with shortened head (brachycephaly). The dog is more likely to have an elongated head (dolichocephaly).
chr10:11072007	C/C	The dog carries two copies of an allele typically associated with floppy ears. The dog is more likely to have floppy than pricked ears.
<i>T</i> c.189C>G (p.Ile63Met)	C/C	The dog does not carry the tested bobtail-causing genetic variant. The dog is most likely long-tailed.

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Test results - Additional disorders found in other breeds - page 1

Blood Disorders - page 1

Disorder	Mode of Inheritance	Result
Bleeding disorder due to P2RY12 defect	Autosomal Recessive	Clear
Canine Cyclic Neutropenia, Cyclic Hematopoiesis, Grey Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	Autosomal Recessive	Clear
Canine Scott Syndrome, (CSS)	Autosomal Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation Gly379Glu	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Airedale Terrier	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso	X-linked Recessive	No call
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Boxer	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Havanaese	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd	X-linked Recessive	Clear
Factor XI Deficiency	Autosomal Dominant (Incomplete Penetrance)	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Hereditary Elliptocytosis		Clear
Hereditary Phosphofructokinase (PFK) Deficiency	Autosomal Recessive	Clear
Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Prekallikrein Deficiency	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Basenji	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Beagle	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 2

Blood Disorders - page 2

Disorder	Mode of Inheritance	Result
Pyruvate Kinase Deficiency; mutation originally found in Pug	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 1	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Kooikerhondje	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Scottish Terrier	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 3

Ocular Disorders - page 1

Disorder	Mode of Inheritance	Result
Canine Multifocal Retinopathy 2, (CMR2); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 3, (CMR3); mutation originally found in Lapponian Herder	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shepherd Dog	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer	Autosomal Recessive	Clear
Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy 2, (crd2); mutation originally found in American Pit Bull Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy, (cord1-PRA / crd4)	Autosomal Recessive (Incomplete Penetrance)	Clear
Cone-Rod Dystrophy, Standard Wirehaired Dachshund, (crd SWD)	Autosomal Recessive	Clear
Generalized Progressive Retinal Atrophy	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)	Autosomal Recessive	Clear
Primary Hereditary Cataract, (PHC); mutation originally found in Australian Shepherd	Autosomal Dominant (Incomplete Penetrance)	Clear
Primary Lens Luxation, (PLL)	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Beagle	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalene	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Basenji	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1, (rcd1); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1a, (rdc1a); mutation originally found in Sloughi	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 4

Ocular Disorders - page 2

Disorder	Mode of Inheritance	Result
Rod-Cone Dysplasia 3, (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1, (XLPRA1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPRA2; Type A PRA)	X-linked Recessive	Clear

Cardiac Disorders

Disorder	Mode of Inheritance	Result
Long QT Syndrome	Autosomal Dominant	Clear

Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Hypothyroidism; mutation originally found in Tenterfield Terrier	Autosomal Recessive	Clear
Congenital Hypothyroidism; mutation originally found in Toy Fox and Rat Terrier	Autosomal Recessive	Clear

Immunological Disorders

Disorder	Mode of Inheritance	Result
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	Clear
Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)	Autosomal Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound	X-linked Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi	X-linked Recessive	Clear

Test results - Additional disorders found in other breeds - page 5

Renal Disorders

Disorder	Mode of Inheritance	Result
Cystinuria Type I-A; mutation originally found in Newfoundland Dog	Autosomal Recessive	Clear
Cystinuria Type II-A; mutation originally found in Australian Cattle Dog	Autosomal Dominant	Clear
Fanconi Syndrome	Autosomal Recessive	Clear
Hyperuricosuria, (HUU)	Autosomal Recessive	Clear
Polycystic Kidney Disease in Bull Terriers, (BTPKD)	Autosomal Dominant	Clear
Primary Hyperoxaluria, (PH); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Protein Losing Nephropathy, (PLN); NPHS1 gene variant		Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN)	X-linked Recessive	Clear
X-Linked Hereditary Nephropathy, (XLHN); mutation originally found in Navasota Dog	X-linked Recessive	Clear

Test results - Additional disorders found in other breeds - page 6

Metabolic Disorders

Disorder	Mode of Inheritance	Result
Glycogen Storage Disease Type II or Pompe's Disease, (GSD II)	Autosomal Recessive	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	Autosomal Recessive	Clear
Glycogen Storage Disease Type Ia, (GSD Ia)	Autosomal Recessive	Clear
Hypocatalasia or Acatalasemia	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Beagle	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Border Collie	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in Dachshund	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in New Zealand Huntaway	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in Brazilian Terrier	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in German Shepherd	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 7

Muscular Disorders

Disorder	Mode of Inheritance	Result
Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)	X-linked Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Great Dane	Autosomal Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Golden Retriever	X-linked Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier	X-linked Recessive	Clear
Muscular Dystrophy, Ullrich-type; mutation originally found in Landseer	Autosomal Recessive	Clear
Myostatin deficiency (Double Muscling, "Bully")	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in Australian Cattle Dog	Autosomal Recessive	Clear
Myotubular Myopathy; mutation originally found in Rottweiler	X-linked Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear

Test results - Additional disorders found in other breeds - page 8

Neurological Disorders - page 1

Disorder	Mode of Inheritance	Result
Alaskan Husky Encephalopathy, (AHE)	Autosomal Recessive	Clear
Bandera's Neonatal Ataxia, (BNAt)	Autosomal Recessive	Clear
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Autosomal Recessive	Clear
Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun	Autosomal Recessive	Clear
Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Fetal Onset Neuroaxonal Dystrophy, (FNAD)	Autosomal Recessive	Clear
Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter	Autosomal Recessive	Clear
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
Hypomyelination; mutation originally found in Weimaraner	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier	Autosomal Recessive	Clear
Lagotto Storage Disease, (LSD)	Autosomal Recessive	Clear
Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)	Autosomal Recessive	Clear
Neonatal Encephalopathy with Seizures, (NEWS)	Autosomal Recessive	Clear
Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Australian Shepherd	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in English Setter	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Autosomal Recessive	Clear
Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 9

Neurological Disorders - page 2

Disorder	Mode of Inheritance	Result
Spinal Dysraphism	Autosomal Recessive	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)	Autosomal Recessive	Clear
Spinocerebellar Ataxia/ Late-Onset Ataxia (SCA, LOA)	Autosomal Recessive	Clear
X-Linked Tremors; mutation originally found in English Springer Spaniel	X-linked Recessive	Clear

Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog	Autosomal Recessive	Clear
Episodic Falling Syndrome, (EFS)	Autosomal Recessive	Clear
Exercise-Induced Collapse, (EIC)	Autosomal Recessive (Incomplete Penetrance)	Clear
GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog	Autosomal Recessive	Clear
GM2 Gangliosidosis, mutation originally found in Japanese Chin	Autosomal Recessive	Clear
GM2 Gangliosidosis; mutation originally found in Toy Poodle	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Terriers	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 10

Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Cleft Palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds	Autosomal Dominant (Incomplete Penetrance)	Clear
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Osteochondrodysplasia; mutation originally found in Miniature Poodle	Autosomal Recessive	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Beagle	Autosomal Dominant	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Dysplasia 2, (SD2)	Autosomal Recessive	Clear
Spondylocostal Dysostosis	Autosomal Recessive	Clear
Van den Ende-Gupta Syndrome, (VDEGS)	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 11

Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa; mutation originally found in Central Asian Ovcharka	Autosomal Recessive	Clear
Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma, (FNEPPK); mutation originally found in Dogue de Bordeaux	Autosomal Recessive	Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in Great Dane	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Ligneous Membranitis	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear

Other Disorders

Disorder	Mode of Inheritance	Result
Amelogenesis Imperfecta, (AI); mutation originally found in Italian Greyhound	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	Autosomal Recessive	Clear
Dental Hypomineralisation; mutation originally found in Border Collie	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Dachshund	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia, (PCD)	Autosomal Recessive	Clear

APPENDIX

Explanation of the results of the tested disorders

Autosomal recessive inheritance (ARI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A dog carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A dog carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

Autosomal dominant inheritance (ADI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A dog carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

X-linked recessive inheritance (X-linked)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female dogs at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Dogs at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.



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