Supplementary data 1. List of primers used in this study

Name	Direction [a]	Position [b]	Sequence (5' to 3')	Usage
mP-1	F	5	GTGCCTGCTGAGAAATCTTACACC	cDNA amplification
mP-2	R	532	AGCAACCTCTTTACCCAGCATG	cDNA amplification
mP-3	F	411	CTCCTCTGCTCTGGAATTCATC	cDNA amplification
mP-4	R	640	GTGATCTTTATCCACCGCACAC	cDNA amplification
mP-5	F	533	ATGCTGGGTAAAGAGGTTGCTG	cDNA amplification
mP-6	R	1261	GGCCAGAGTCTCGAAATCAATC	cDNA amplification
mP-7	F	1261	GATTGATTTCGAGACTCTGGCC	cDNA amplification
mP-8	R	1968	ACTTGACAAGCAGACTCCTGTCTG	cDNA amplification
mP-9	F	1090	AAGCGTTGAAGCACAAGTGG	cDNA amplification
mP-10	R	1506	TGCAATGAGGACTTGTCTCG	cDNA amplification
mP-11	F	1506	CGAGACAAGTCCTCATTGCAGAAG	cDNA amplification
mP-12	R	2333	CGTAGCAGTAAATGGGATGTTGTC	cDNA amplification
mP-13	F	2333	GACAACATCCCATTTACTGCTACG	cDNA amplification
mP-14	R	2602	TCCCACCACAATGTGAGCAATC	cDNA amplification
mP-15	F	2173	AGCACTCTTTGTGCTGATGG	cDNA amplification
mP-16	R	2351	GGAGTACAGGAATCATCGTAGC	cDNA amplification
mP-17	F	85592	GGACAAAACAGTGGTGCTTG	Deletion region amplification
mP-18	R	91472	GAGCACAAGCTGCCTCAAG	Deletion region amplification
mP-19	R	91805	CTCCGTGGGAAAGGATACAC	Deletion region amplification
mP-20	F		TCCTGAGTTGGAGGAGAACAAG	Promoter region amplificatio
mP-21	R		TACCTGATGACTCGCTGATCTC	Promoter region amplificatio
mP-22	F		CTAATCTGTATGGAGGCACTGG	Promoter region amplificatio
mP-23	R		CAAGCCTTCATAGGTGGCATAG	Promoter region amplificatio
mP-24	F		ACCTGATAAGGCTTGGCTGAG	Promoter region amplificatio
mP-25	R		AGAGACACACACAGGTACAGAAGC	Promoter region amplificatio
Oca2	F	530	TGCATGCTGGGTAAAGAGGTT	RT-qPCR
	R	595	TGCAAGATCCCGTTTCTCTGA	RT-qPCR
Actb	F		GGCTGTATTCCCCTCCATCG	RT-qPCR
	R		CCAGTTGGTAACAATGCCATGT	RT-qPCR
B2m	F		TTCTGGTGCTTGTCTCACTGA	RT-qPCR
	R		CAGTATGTTCGGCTTCCCATTC	RT-qPCR

[a] F : forward primer; R: reverse primer. [b] The position of 5' end of the primer to the mouse *Oca2* cDNA (NM_021879.2) for mP-1 to mP-16 and to the mouse *Oca2* genome (NC_000073.5) for mP-17 to mP-25

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Wt      1 MRLENKDIRLASAVLEVELHQTSALSVPTCPDPGRLLTVKPATSNYKLGQADPCIPYAGEAAGKSVCVPEHTEFGSFLVKGSSSLKDLSFKEDTPLLWNS
p-cas     MRLENKDIRLASAVLEVELHQTSALSVPTCPDPGRLLTVKPATSNYKLGQADPCIPYAGEAAGKSVCVPEHTEFGSFLVKGSSSLKDLSFKEDTPLLWNS

Wt    101 SQKKRSQLMPVHHPEFIATEGSWENGLTAWEQKCMLGKEVADLSALASSEKRDLAGSVHLRAQVSKLGCCVRWIKITGLFVFVVLCSILFSLYPDQGKFW
p-cas     SQKKRSQLMPVHHPEFIATEGSWENGLTAWEQKCMLGKEVADLSALASSEKRDLAGSVHLRAQVSKLGCCVRWIKITGLFVFVVLCSILFSLYPDQGKFW

Wt    201 QLLAVSPLENYSVNLSGHADSMILQLDLAGALMAGGPSGSGKEEHVVVVVTQTDAAGNRRRRPQQLTYNWTVLLNPRSEHVVVSRTFEIVSREAVSISIQ
p-cas     QLLAVSPLENYSVNLSGHADSMILQLDLAGALMAGGPSGSGKEEHVVVVVTQTDAAGNRRRRPQQVTYNWTVLLNPRSEHVVMSRTFEIVSREAVFISIQ

Wt    301 ASLQQTRLVPLLLAHQFLGASVEAQVASAVAILAGVYTLIIFEIVHRTLAAMLGALAALAALAVVGDRPSLTHVVEWIDFETLALLFGMMILVAVFSETG
p-cas     ASLQQTRLVPLLLAHQFLGASVEAQVASAVAILAGVYTLIIFEIVHRTLAAMLGALAALAALAVVGDRPSLTHVVEWIDFETLALLFGMMILVAVFSETG

Wt    401 FFDYCAVKAYQLSRGRVWAMIFMLCLMAAILSAFLDNVTTMLLFTPVTIRLCEVLNLDPRQVLIAEVIFTNIGGAATAIGDPPNVIIVSNQELRKMGLDF
p-cas     FFDYCAVKAYQLSRGRVWAMIFMLCLMAAILSAFLDNVTTMLLFTPVTIRLCEVLNLDPRQVLIAEVIFTNIGGAATAIGDPPNVIIVSNQELRKMANFT

Wt    501 AGFTAHMFLGICLVLLVSFPLLRLLYWNKKLYNKEPSEIVELKHEIHVWRLTAQRISPASREETAVRGLLLEKVLALEHLLAQRLHTFHRQISQEDKNWE
p-cas     GRQKLGDQYSRATKKAQDFRQESACQVPDGAGICHLHVLSQLLCPWHSS*----------------------------------------------------

Wt    601 TNIQELQRKHRISDRSLLVKCLTVLGFVISMFFLNSFVPGIHLDLGWIAILGAIWLLILADIHDFEIILHRVEWATLLFFAALFVLMEALTHLHLVEYVG
p-cas     ----------------------------------------------------------------------------------------------------

Wt    701 EQTALLIKMVPEDQRFAAAIVLIVWVSALASSLIDNIPFTATMIPVLLNLSQDPEISLPALPLMYALALGACLGGNGTLIGASTNVVCAGIAEKHGYGFS
p-cas     ----------------------------------------------------------------------------------------------------

Wt    801 FMEFFRLGFPVMLMSCTIGMCYLLIAHIVVGWN
p-cas     ---------------------------------
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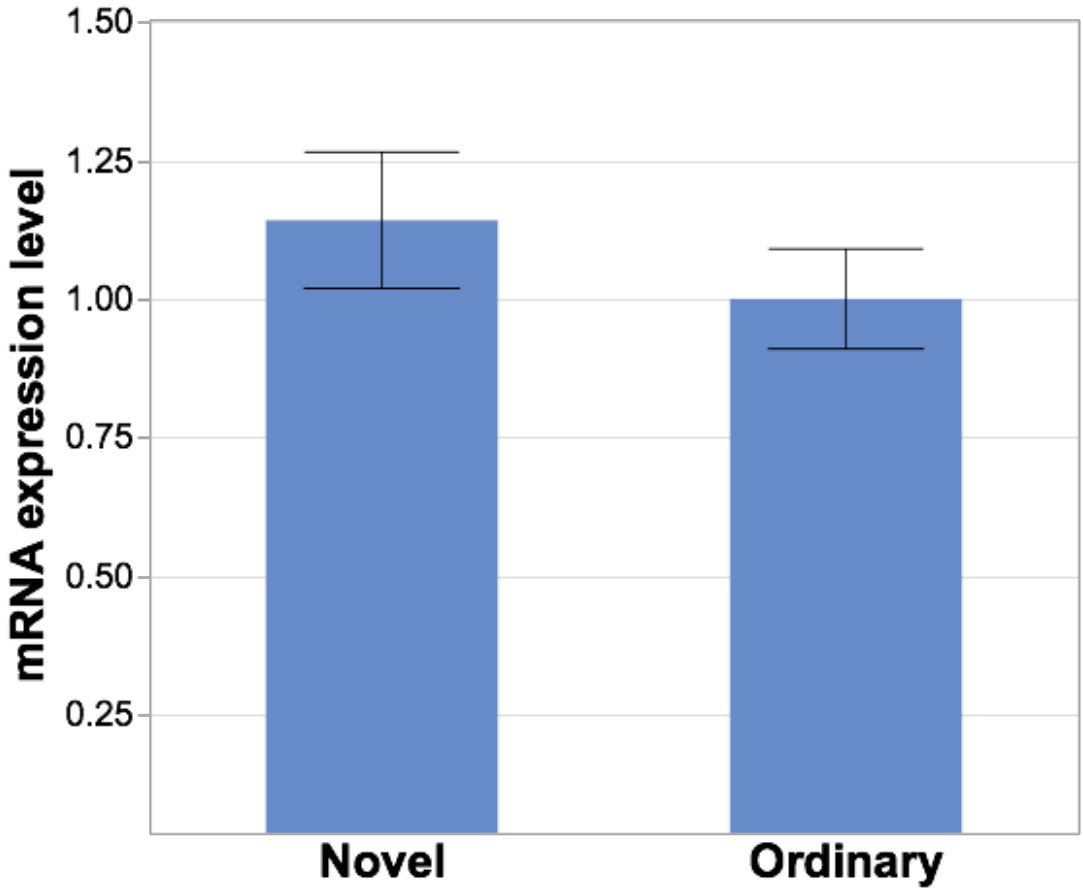
Supplementary data 2. Comparison of amino acid sequences deduced from the cDNA sequences between the wild-type *Oca2*$^{+}$ (Wt) allele and the mutant *Oca2*$^{p\text{-}cas}$ (p-cas) allele. Amino acid substitutions are colored in red. The total number of amino acids in the wild-type allele was 833, whereas that of the mutant allele was only 549 due to complete deletion of exons 15 and 16. The *Oca2*$^{p\text{-}cas}$ cDNA sequence was deposited in the DDBJ/EMBL/GenBank databases with the accession number AB716353. *: creation of a new stop codon; -: deletion.

Supplementary data 3. Comparison of a 968-bp sequence of the promoter regions between the wild-type *Oca2*$^{+}$ (Wt) and mutant *Oca2*$^{p\text{-}cas}$ (p-cas)

alleles. The nucleotide G colored in blue indicates the transcriptional start site (+1) of exon 1. The three nucleotides ATG colored in blue indicate an initiation codon coding for methionine residue (Met). Putative DNA binding domains searched for by the web-based software program TFSEARCH (http://www.cbrc.jp/research/db/TFSEARCH.html) are shown by the boxed nucleotides labeled with letters a to d. a: OSE2 (osteoblast-specific cis-acting element 2) motif; b: RUNX1 (runt-related transcription factor 1) binding motif; c: zinc finger motif; d: homeodomain motif. Nucleotide substitutions are colored in red. The *Oca2*$^{p\text{-}cas}$ promoter sequence was deposited in the DDBJ/EMBL/GenBank databases with the accession number AB716354. -: deletion.



Supplementary data 4. Comparison of *Oca2*$^{p\text{-}cas}$ expression levels in eyes between novel and ordinary mutant mice. The expression levels were determined by a quantitative relative standard curve method of RT-qPCR and normalized to two endogeous control genes of *Actb* and *B2m*. Data are expressed as the mean and one standard error for three mice at about 4 months of age. There was no significant difference in expression level between the two types of mutant mice (t-test, P=0.404).