

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 | GATTGATTTTCGAGACTCTGGCC | 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 amplification |
| mP-21 | R | | TACCTGATGACTCGCTGATCTC | Promoter region amplification |
| mP-22 | F | | CTAATCTGTATGGAGGCACTGG | Promoter region amplification |
| mP-23 | R | | CAAGCCTTCATAGGTGGCATAG | Promoter region amplification |
| mP-24 | F | | ACCTGATAAGGCTTGGCTGAG | Promoter region amplification |
| mP-25 | R | | AGAGACACACAGGTACAGAAGC | Promoter region amplification |
| <i>Oca2</i> | F | 530 | TGCATGCTGGGTAAAGAGGTT | RT-qPCR |
| | R | 595 | TGCAAGATCCCGTTTCTCTGA | RT-qPCR |
| <i>Actb</i> | F | | GGCTGTATCCCCTCCATCG | RT-qPCR |
| | R | | CCAGTTGGTAACAATGCCATGT | RT-qPCR |
| <i>B2m</i> | F | | TTCTGGTGCTTGTCTCACTGA | RT-qPCR |
| | R | | CAGTATGTTCCGCTTCCCATTC | 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

Wt 1 MRLNKDIRLASAVLEVELHQTSALSVPTCPDPGRLLTVKPATSNYKLGQADPCIPYAGEAAGKSVCVPEHTEFGSFLVKGSSSLKDLSFKEDTPLLWNS
p-cas MRLNKDIRLASAVLEVELHQTSALSVPTCPDPGRLLTVKPATSNYKLGQADPCIPYAGEAAGKSVCVPEHTEFGSFLVKGSSSLKDLSFKEDTPLLWNS

Wt 101 SQKKRSQLMPVHHPEFIATEGSWENGLTAWQKCMKGKEVADLSALASSEKRDLAGSVHLRAQVSKLGCCVRWIKITGLFVFFVLCILFSLYPDQGKFW
p-cas SQKKRSQLMPVHHPEFIATEGSWENGLTAWQKCMKGKEVADLSALASSEKRDLAGSVHLRAQVSKLGCCVRWIKITGLFVFFVLCILFSLYPDQGKFW

Wt 201 QLLAVSPLENYSVNLSGHADSMILQLDLAGALMAGGPSGSGKEEHVVVVVTQTDAAGNRRRRPQQLTYNWTVLLNPRSEHVVSRTFEIVSREAVSISIQ
p-cas QLLAVSPLENYSVNLSGHADSMILQLDLAGALMAGGPSGSGKEEHVVVVVTQTDAAGNRRRRPQQVTYNWTVLLNPRSEHVMSRTFEIVSREAVFISIQ

Wt 301 ASLQQTRLVPLLLAHQFLGASVEAQVASAVAILAGVYTLIIFEIVHRTLAAMLGALAALAALAVGDRPSLTHVVEWIDFETLALLFGMMILVAVFSETG
p-cas ASLQQTRLVPLLLAHQFLGASVEAQVASAVAILAGVYTLIIFEIVHRTLAAMLGALAALAALAVGDRPSLTHVVEWIDFETLALLFGMMILVAVFSETG

Wt 401 FFDYCAVKAYQLSRGRVWAMIFMLCLMAAILSAFLDNVTMMLFTPVTIRLCEVLNLDPRQVLI AEVIFTNIGGAATAIGDPPNVIIVSNQELRKMGLDF
p-cas FFDYCAVKAYQLSRGRVWAMIFMLCLMAAILSAFLDNVTMMLFTPVTIRLCEVLNLDPRQVLI AEVIFTNIGGAATAIGDPPNVIIVSNQELRKM ANFT

Wt 501 AGFTAHMFLGICLVLLVSFPLLRLLYWNKKLYNKEPSEIVELKHEIHVWRLTAQRISPASREETA VRGLLLEKVLALHLLAQRLHTFHRQISQEDKNWE
p-cas GRQKLGQYSRATKKAQDFRQESACQVPDGAGICHHLVLSQLLCPWHSS*-----

Wt 601 TNIQELQRKHRSRSLLVKCLTVLGFVISMFFLNSFVPGIHLDLGWIAILGAIWLLILADIHDFEILHRVEWATLLFFAALFVLMEALTHLHLVEYVG
p-cas -----

Wt 701 EQTALLIKMPEDQRFAAAIVLIVVWSALASSLIDNIPFTATMIPVLLNLSQDPEISLPALPLMYALALGACLGNGTLIGASTNVVCAGIAEKHGYGFS
p-cas -----

Wt 801 FMEFFRLGFPVMLMSCTIGMICYLLIAHIVVGWN
p-cas -----

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-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-cas} cDNA sequence was deposited in the DDBJ/EMBL/GenBank databases with the accession number AB716353. *: creation of a new stop codon; -: deletion.

Wt -968 GCGGGGGGGGTTGAGCAAAATGGCTCAGCAAGTAAAGGCACTTGCTGCGGAACCCAGTGACCTGACTTTGGTCACTGGAAACCACACGCTAGAAGGAGA
p-cas GGGGGTGGGGGTGAGCAAAATGGCTCAGCAAGTAAAGGCACTTGCTGCGGAACCCAGTGACCTGACTTTGGTCACTGGAAACCACACGCTATAAGGAGA

Wt -868 AAAGTAACTCCGCGAGTTGTCTCCTGTGGCATGCATGAA-----CACTCA
p-cas AAAGTAACTCTGCGAGTTGTCTCCCGTGGCATGCATGAAACTCA

Wt -768 TACAAAATAGTTTAAAAATTTAAAAGTCTGTGAATGATGGGGGAGTGGAAAGCAGCTCCCTTGGCTGTTACAGTGACTACTTTAACCCTTTGCTAATCTG
p-cas TACAAAATAGTTTAAAAATTTAAAAGTCTGTGAATGATGGGGGAGTGGAAAGCAGCTCCCTTGGCTGTTACAGTGACTACTTTAACCCTTTGCTAATCTG

Wt -668 TATGGAGGCACTGGGACTGCTGGATTCTGGCACTGTGTCATCCTGTAGTTCATGTCTGCATGTATGATATGTCTTATGTGCCTGCTTCTGTACCTGTG
p-cas TATGGAGGCACTGGGACTGCTGGATTCTGGCACTGTGTCATCCTGTAGTTTATGTCTGCATGTATGATATGTCTTATGTGCCTGCTTCTGTACCTGTG

Wt -568 TGTGTCTCTCTCTTTGTGTATGACTTCATGAGTGTGGTGGTCTGTGCATGTTGTGTTTATAACTGCATATCCCTCCTTCTATATGGGCTTTTGGGTAT
p-cas TGTGTCTCTCTCTGTGTGTATGACTTCATGAGTGCATTGGGTCTGTGCATTTGTGTTTGTAACTGCATATCCCTCCTTCTATATGGGCTTTTGGGTAT

Wt -468 TTGTACAGTTTGTGTATTTCCATGTAATTTATCATTCTGTGAGCAGTCTTTGCGTATTTATTCATGGTCATGGTGTGTGGTATGATGGAGCTTTCCTT
p-cas TTGTACAGTTTGTGTGTTTCCATGTAATTTATCATTCTGTGAGCAGTCTTTGCGTATTTATTCATGGTCATGCGTGTGCGGTTTGATGGAGCTTTCCTT

Wt -368 GGATGTAACCTTGCCACACTCCTGAGTTGGAGGAGACAAGATAATGAGAAAGTTTATCCTCTTCAGTAGACAGTTCTTACAATGCAGAACAATTTATA
p-cas GGATGTAACCTTGCCACACTCCTGAGTTGGAGGAGACAAGATAATGAGAAAGTTTATCCTCTTCAGTAGACAGTTCTTACAATGCAGAACAATTTATA

Wt -268 TTTCTCAGTTCTTGCCTTAAGGTA AAAATCTGCCACGTCCTATGCCACCTATGAAGGCTTGAGCTTGCTACACATGGCTGGAATTTTCCAAAAGCCTGGC
p-cas TTTCTCAGTTCTTGCCTTAAGGTA AAAATCTGCCACGTCCTATGCCACCTATGAAGGCTTGAGCTTGCTACACATGGCTGGAATTTTCCAAAAGCCTGGC

Wt -168 TTCTACAGCAAGGCTCTCTGTTTTCCAGCTGAGGCTCCTGCATGCTGCTGGACCATTCTCAAGAGGGGGCTGGAGCCACTGAGCCTTCAACTTTGTGAA
p-cas CACTGCAGCAAGGCTCTCTGTTTTCCAGCTGAGGCTCCTGCATGCTGCTGGACCATTCTCAAGAGGGGGCTGGAGCCACTGAGCCTTCAACTTTGTGAA

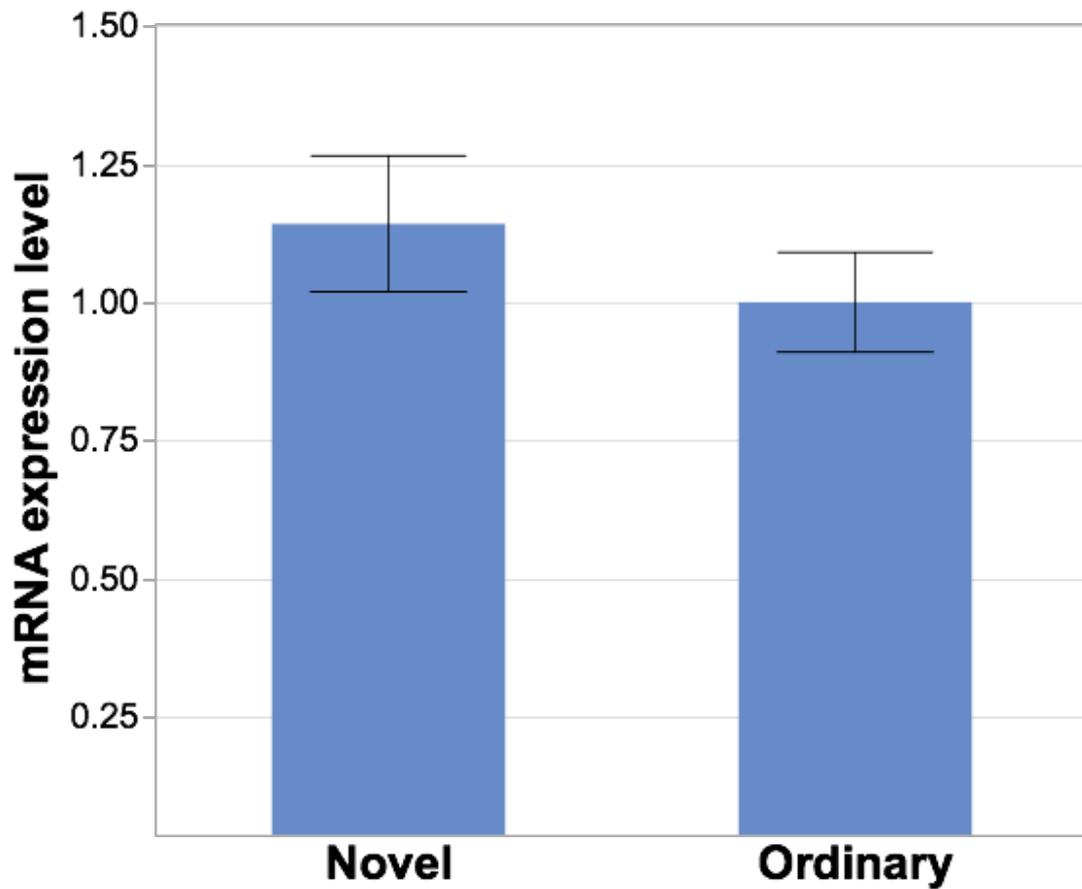
Wt -68 GGTCTGTGGGCGTGGCCAGCACAGGGTGCAGTGAGGAGCACAAGCTATCCAACCCTCCCTCTGGGGCTGCAAGTGCCTGCTGAGAAATCTTACACCAGGG
p-cas GGTCTGTGGGTGTGGCCAGCAGAGGGTGCAGTAAGGAGCACAAGCTATCCAACCCTCCCTCTGGGGCTGCAAGTGCCTGCTGAGAAATCTTACACCAGGG

Wt 33 TTGTGCTCCATCCACGACTCAGAGCCTTTGGATCTGGACACTAGACTTCACTGCTGGAGAGAGATCAGCGAGTCATCAGACAGATCAGCAACGGGGACATG
p-cas TTGTGCTCCATCCACGACTCAGAGCCTGTGGATCTGGACACTAGACTTCACTGCTGGAGAGAGATCAGCGAGTCATCAGACAGATCAGCAACGGGGACATG

Met

Supplementary data 3. Comparison of a 968-bp sequence of the promoter regions between the wild-type *Oca2*⁺ (Wt) and mutant *Oca2*^{p-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-cas} promoter sequence was deposited in the DDBJ/EMBL/GenBank databases with the accession number AB716354. -: deletion.



Supplementary data 4. Comparison of *Oca2^{p-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 endogenous 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, $P=0.404$).