## Kim M Summers

## List of Publications by Year in descending order

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| 1 | A promoter-level mammalian expression atlas. Nature, 2014, 507, 462-470. | 13.7 | 1,838 |
| :---: | :---: | :---: | :---: |
| 2 | Microglial brain regionâ^'dependent diversity and selective regional sensitivities to aging. Nature Neuroscience, 2016, 19, 504-516. | 7.1 | 919 |
| 3 | Transcribed enhancers lead waves of coordinated transcription in transitioning mammalian cells. Science, 2015, 347, 1010-1014. | 6.0 | 517 |
| 4 | An integrated expression atlas of miRNAs and their promoters in human and mouse. Nature Biotechnology, 2017, 35, 872-878. | 9.4 | 456 |
| 5 | Abnormal Extracellular Matrix Protein Transport Associated With Increased Apoptosis of Vascular Smooth Muscle Cells in Marfan Syndrome and Bicuspid Aortic Valve Thoracic Aortic Aneurysm. Circulation, 2003, 108, 32911-334. | 1.6 | 224 |
| 6 | A high resolution atlas of gene expression in the domestic sheep (Ovis aries). PLoS Genetics, 2017, 13, el006997. | 1.5 | 210 |
| 7 | A gene expression atlas of the domestic pig. BMC Biology, 2012, 10, 90. | 1.7 | 199 |

$8 \quad$ FANTOM5 CAGE profiles of human and mouse samples. Scientific Data, 2017, 4, 170112.
2.4

195

| 9 | Deletion of a Csf1r enhancer selectively impacts CSF1R expression and development of tissue macrophage populations. Nature Communications, 2019, 10, 3215. | 5.8 | 191 |
| :---: | :---: | :---: | :---: |
| 10 | Analysis of the human monocyte-derived macrophage transcriptome and response to lipopolysaccharide provides new insights into genetic aetiology of inflammatory bowel disease. PLoS Genetics, 2017, 13, e1006641. | 1.5 | 161 |
| 11 | Identification of homozygous hemochromatosis subjects by measurement of hepatic iron index. Hepatology, 1990, 12, 20-25. | 3.6 | 148 |
| 12 | Differentiation Between Heterozygotes and Homozygotes in Genetic Hemochromatosis by Means of a Histological Hepatic Iron Index: A Study of 192 Cases. Hepatology, 1993, 17, 30-34. | 3.6 | 147 |
| 13 | A comprehensive biomedical variant catalogue based on whole genome sequences of 582 dogs and eight wolves. Animal Genetics, 2019, 50, 695-704. | 0.6 | 138 |

14 Regional mapping panel for human chromosome 17: Application to neurofibromatosis type 1 . Genomics, 1987, 1, 374-381.
1.3

126
15 A Deletion in the Canine POMC Gene Is Associated with Weight and Appetite in Obesity-Prone Labrador Retriever Dogs. Cell Metabolism, 2016, 23, 893-900.

Pleiotropic Impacts of Macrophage and Microglial Deficiency on Development in Rats with Targeted Mutation of the <i>Csflr</i> Locus. Journal of Immunology, 2018, 201, 2683-2699.
19 Large animal models of cardiovascular disease. Cell Biochemistry and Function, 2016, 34, 113-132.

1.4

105

20 Phenotypic impacts of CSF1R deficiencies in humans and model organisms. Journal of Leukocyte
1.5

97
Biology, 2020, 107, 205-219.

21 Biology of Eye Pigmentation in Insects. Advances in Insect Physiology, 1982, , 119-166.
1.1

95

Network analysis of transcriptomic diversity amongst resident tissue macrophages and dendritic cells in the mouse mononuclear phagocyte system. PLoS Biology, 2020, 18, e3000859.
2.6

Structure and function of the mammalian fibrillin gene family: Implications for human connective
$0.5 \quad 89$
tissue diseases. Molecular Genetics and Metabolism, 2012, 107, 635-647.

Combination of novel and public RNA-seq datasets to generate an mRNA expression atlas for the
domestic chicken. BMC Genomics, 2018, 19, 594.
1.2

86

25 Molecular genetics of long QT syndrome. Molecular Cenetics and Metabolism, 2010, 101, 1-8.
0.5

75

Developmental patterns of 3-hydroxykynurenine accumulation in white and various other eye color mutants of Drosophila melanogaster. Biochemical Genetics, 1977, 15, 1049-1059.
$27 \quad$ Multilocus analysis of the fragile X syndrome. Human Genetics, 1988, 78, 201-205.
1.8

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29 Population structure and genetic heterogeneity in popular dog breeds in the UK. Veterinary Journal,
2013, 196, 92-97.
29 2013, 196, 92-97.
```

0.6

55

Macrophage colony-stimulating factor (CSF1) controls monocyte production and maturation and the
30 steady-state size of the liver in pigs. American Journal of Physiology - Renal Physiology, 2016, 311,
1.6

55
G533-G547.
Molecular dissection of a contiguous gene syndrome: frequent submicroscopic deletions,
31 evolutionarily conserved sequences, and a hypomethylated "island" in the Miller-Dieker chromosome
$3.3 \quad 53$
region.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 5136-5140.

The Value of Screening in Siblings of Patients with Abdominal Aortic Aneurysm. European Journal of

Expression of mesenchyme-specific gene signatures by follicular dendritic cells: insights from the
33 Expression of mesenchyme-specific gene signatures by follicular dendritic cells: insights from the
2.0

50

A Gene Expression Atlas of the Domestic Water Buffalo (Bubalus bubalis). Frontiers in Genetics, 2019,
10, 668.
1.1

49

## 35 Characterization of Subpopulations of Chicken Mononuclear Phagocytes That Express TIM4 and CSF1R. <br> Journal of Immunology, 2019, 202, 1186-1199.

0.4

47

CSF1R-dependent macrophages control postnatal somatic growth and organ maturation. PLoS
Genetics, 2021, 17, e1009605.

Functional Annotation of the Transcriptome of the Pig, Sus scrofa, Based Upon Network Analysis of an RNAseq Transcriptional Atlas. Frontiers in Genetics, 2019, 10, 1355.

Alpha-thalassemia in Papua New Guinea. Human Genetics, 1986, 74, 432-437.
1.8

Relationship between genotype and phenotype in monogenic diseases: Relevance to polygenic diseases. ,
1996, 7, 283-293.

Transcriptional switching in macrophages associated with the peritoneal foreign body response.
Immunology and Cell Biology, 2014, 92, 518-526.

Integration of quantitated expression estimates from polyA-selected and rRNA-depleted RNA-seq
libraries. BMC Bioinformatics, 2017, 18, 301.

Co-expression of FBN1 with mesenchyme-specific genes in mouse cell lines: implications for phenotypic
variability in Marfan syndrome. European Journal of Human Genetics, 2010, 18, 1209-1215.

Transcriptional profiling of the human fibrillin/LTBP gene family, key regulators of mesenchymal cell functions. Molecular Genetics and Metabolism, 2014, 112, 73-83.

Fragmentation of tissue-resident macrophages during isolation confounds analysis of single-cell
preparations from mouse hematopoietic tissues. Cell Reports, 2021, 37, 110058.

Concordance of iron storage in siblings with genetic hemochromatosis: Evidence for a predominantly genetic effect on iron storage. Hepatology, 1993, 17, 833-837.

47 Transcriptional Regulation and Macrophage Differentiation. Microbiology Spectrum, 2016, 4, .
1.2

35

Identification of the macrophage-specific promoter signature in FANTOM5 mouse embryo developmental time course data. Journal of Leukocyte Biology, 2017, 102, 1081-1092.
1.5

35

49 Challenges in the diagnosis of Marfan syndrome. Medical Journal of Australia, 2006, 184, 627-631.
0.8

33

Anterior segment mesenchymal dysgenesis in a large Australian family is associated with the recurrent 17 bp duplication in PITX3. Molecular Vision, 2008, 14, 2010-5.

Highly polymorphic locus D15S24 (CMW-1) maps to 15pter-q13. [HGM9 provisional no. D15S24]. Nucleic
Acids Research, 1988, 16, 8740-8740.

Comparative transcriptome analysis of equine alveolar macrophages. Equine Veterinary Journal, 2017, 49, 375-382.

Physical and genetic mapping of the telomeric major histocompatibility complex region in man and relevance to the primary hemochromatosis gene (HFE). Genomics, 1992, 14, 232-240.
1.3

29

Analysis of homozygous and heterozygous Csflr knockout in the rat as a model for understanding
56 microglial function in brain development and the impacts of human CSF1R mutations. Neurobiology of
2.1

29
Disease, 2021, 151, 105268.

Production of a recombinant form of early pregnancy factor that can prolong allogeneic skin graft
survival time in rats. Immunology and Cell Biology, 2000, 78, 603-607.
1.0

Dogslife: A web-based longitudinal study of Labrador Retriever health in the UK. BMC Veterinary
Research, 2013, 9, 13.

Platelet monoamine oxidase: specific activity and turnover number in headache. Clinica Chimica Acta,
1982, 121, 139-146.

RENIN AND ANGIOTENSIN-CONVERTING ENZYME GENOTYPES IN PATIENTS WITH ESSENTIAL HYPERTENSION
60 AND LEFT VENTRICULAR HYPERTROPHY. Clinical and Experimental Pharmacology and Physiology, 1994, 21,
$0.9 \quad 25$ 207-210.

Angiotensin I Converting Enzyme Gene Cosegregates with Blood Pressure and Heart Weight in F2
61 Progeny Derived from Spontaneously Hypertensive and Normotensive Wistar-Kyoto Rats. Clinical and
$0.5 \quad 25$
Experimental Hypertension, 1996, 18, 753-771.
62 The Transcriptional Network That Controls Growth Arrest and Macrophage Differentiation in the
Human Myeloid Leukemia Cell Line THP-1. Frontiers in Cell and Developmental Biology, 2020, 8, 498.

Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in
osteogenesis imperfecta and Marfan syndrome. BoneKEy Reports, 2013, 2, 456.

Xanthommatin biosynthesis in wild-type and mutant strains of the Australian sheep blowfly Lucilia
cuprina. Biochemical Genetics, 1978, 16, 1153-1163.

The Human Early Pregnancy Factor/Chaperonin 10 Gene Family. Biochemical and Molecular Medicine,
$65 \quad \begin{aligned} & \text { The Human Early } \\ & 1996,58,52-58 .\end{aligned}$
1.5

23

Genome-wide analysis of mitral valve disease in Cavalier King Charles Spaniels. Veterinary Journal,
2012, 193, 283-286.
0.6

23

Analysis of gene expression in the nervous system identifies key genes and novel candidates for health and disease. Neurogenetics, 2017, 18, 81-95.

Autosomal dominant cataracts and Peters anomaly in a large Australian family. Clinical Genetics, 1999, 55, 240-247.
1.0

21

## ANGIOTENSIN-CONVERTING ENZYME AND ANGIOTENSINOGEN GENES IN PATTERNS OF LEFT VENTRICULAR

69 HYPERTROPHY AND IN DIASTOLIC DYSFUNCTION. Clinical and Experimental Pharmacology and
0.9

20
Physiology, 1995, 22, 438-440.
Histopathology and fibrillin-1 distribution in severe early onset Marfan syndrome. American Journal

Experimental and bioinformatic characterisation of the promoter region of the Marfan syndrome gene, FBN1. Genomics, 2009, 94, 233-240.
73 Dogslife: A cohort study of Labrador Retrievers in the UK. Preventive Veterinary Medicine, 2015, 122,
426-435.
GENETIC VARIANTS OF PROTEINS FROM THE RENIN ANGIOTENSIN SYSTEM ARE ASSOCIATED WITH PRESSURE
$0.9 \quad 14$

Recent developments in the diagnosis of Marfan syndrome and related disorders. Medical Journal of

92 Validity of Internet-Based Longitudinal Study Data: The Elephant in the Virtual Room. Journal of
99 Macrophages form erythropoietic niches and regulate iron homeostasis to adapt erythropoiesis in
?1-ANTITRYPSIN DEFICIENCY ALLELES AND BLOOD PRESSURE IN AN AUSTRALIAN POPULATION. Clinical and
101 Experimental Pharmacology and Physiology, 1996, 23, 600-601.
0.9 ..... 8Role of macrophages and phagocytes in orchestrating normal and pathologic hematopoietic niches.
103 Albumin ? vitamin D-binding protein haplotypes in Asian-Pacific populations. Human Genetics, 1990, 85, 89-97.

Familial muscular ventricular septal defects and aneurysms of the muscular interventricular septum.

ANALYSIS OF LINKAGE OF THE ACE LOCUS WITH MEASURES OF CARDIAC HYPERTROPHY IN THE
114 SPONTANEOUSLY HYPERTENSIVE RAT. Clinical and Experimental Pharmacology and Physiology, 1996, 23, ..... 0.9
597-599.
115 Mapping and characterization of the eukaryotic early pregnancy factor/chaperonin 10 gene family. Somatic Cell and Molecular Genetics, 1998, 24, 315-326.$0.7 \quad 5$
116 Compound heterozygous mutations in $\langle\mathrm{i}\rangle \mathrm{FBN} 1</ \mathrm{i}\rangle$ in a large family with Marfan syndrome. MolecularGenetics \& Genomic Medicine, 2020, 8, el116.
0.65
117 Improving the resolution of canine genomeâ€wide association studies using genotype imputation: A
study of two breeds. Animal Genetics, 2021, 52, 703-713.0.65
Fine mapping of a human chromosome 6 ferritin heavy chain pseudogene: relevance to
1.8 ..... 4
118 haemochromatosis. Human Genetics, 1991, 88, 175-8.Mutations at <i>KCNQ1</i> and an unknown locus cause long QT syndrome in a large Australian119 family: Implications for genetic testing. American Journal of Medical Genetics, Part A, 2010, 152A,0.7613-621.
120 Visualization and analysis of RNA-Seq assembly graphs. Nucleic Acids Research, 2019, 47, 7262-7275.6.54Concordance of iron storage in siblings with genetic hemochromatosis: Evidence for a predominantly3.6genetic effect on iron storage,. Hepatology, 1993, 17, 833-837.Generation and network analysis of an RNA-seq transcriptional atlas for the rat. NAR Genomics and1.5Bioinformatics, 2022, 4, lqac017.Regulation of the production of granulocyte-macrophage colony-stimulating factor bymacrophage-like tumour cell lines. FEBS Letters, 1985, 180, 271-274.1.3Limited genetic divergence between dog breeds from geographically isolated countries. VeterinaryRecord, 2014, 175, 562-562.
in Brief, 2016, 8, 851-857.
Relationship between genotype and phenotype in monogenic diseases: Relevance to polygenic diseases.
Human Mutation, 1996, 7, 283-293.

Applications of molecular genetics to gastrointestinal and liver diseases. I. Technical approaches. Journal of Gastroenterology and Hepatology (Australia), 1989, 4, 183-193.

Is determination of the hepatic iron index of diagnostic value in patients with thalassemia minor and chronic alcoholic liver disease?. Hepatology, 1991, 14, 959-960.

ASSOCIATION ANALYSIS OF SIX CANDIDATE GENES IN A SAMPLE OF AUSTRALIAN HYPERTENSIVE PATIENTS. Clinical and Experimental Pharmacology and Physiology, 1997, 24, 454-456.

Microdeletion of 9 q 22.3 : A patient with minimal deletion size associated with a severe phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 2070-2083.

CRISPR-Cas9 Editing of Human Histone Deubiquitinase Gene USP16 in Human Monocytic Leukemia Cell
Line THP-1. Frontiers in Cell and Developmental Biology, 2021, 9, 679544.

Genetic distance analysis using DNA polymorphisms in the $\hat{I}_{ \pm-g l o b i n ~ g e n e ~ c l u s t e r . ~ A n n a l s ~ o f ~ H u m a n ~}^{\text {g }}$ Biology, 1987, 14, 393-404.

Identifying susceptibility to inflammatory bowel diseases: A candidate gene approach, genomeấwide association studies, or both?. Journal of Gastroenterology and Hepatology (Australia), 2008, 23, 6-7.

Transcriptional Regulation and Macrophage Differentiation. , 2017, , 117-139.

Canine reference genome accuracy impacts variant calling: Lessons learned from investigating embryonic lethal variants. Animal Genetics, 2022, 53, 706-708.
? $i i^{1 / 2}$ - and $?_{i} i^{1 / 2}$ - Thalassemia in a Thai family: unusually mild homozygous $?_{i} i^{1 / 2}$-thalassemia without ?-globin gene deletion. Human Genetics, 1985, 69, 375-377.

Applications of molecular genetics to gastrointestinal and liver diseases. II. Clinical relevance.
Journal of Gastroenterology and Hepatology (Australia), 1989, 4, 273-281.

Genetic heterogeneity in Wilson's disease. Journal of Gastroenterology and Hepatology (Australia), 1990, 5, 697-699.
â $€^{\sim}$ Dogslifeâ $€^{\text {TM }}$ research study. Veterinary Record, 2010, 167, 146-146.

16â€...Investigating calcific aortic valve disease using novel immortalised sheep and rat valve interstitial cell lines. , 2017, , .

8â€...Generating a genomic-wide transcriptomic atlas of the mammalian cardiovascular system. , 2017, , .

Identification of Pathological FBN1 Variants Is Not Straightforward. Circulation Genomic and
Precision Medicine, 2018, 11, e002168.

Clinical and Echocardiographic Findings in an Aged Population of Cavalier King Charles Spaniels.
Animals, 2021, 11, 949.


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