Lecture 6:

Analytics and metrics:

Journal, article, and authors

Course 501

Writing and Communication Skills

AIMS

- Learn about the analytics metrics of journals, articles, and authors.
- Use the analytics you learn to:
    - Identifying good references.
    - Refine your previous literature search.

Journal analytics and metrics

FUNDAMENTAL CONCEPTS IN GENETICS

Linkage disequilibrium — understanding the evolutionary past and mapping the medical future

Montgomery Slatkin

Abstract | Linkage disequilibrium — the nonrandom association of alleles at different loci — is a sensitive indicator of the population genetic forces that structure a genome. Because of the explosive growth of methods for assessing genetic variation at a fine scale, evolutionary biologists and human geneticists are increasingly exploiting linkage disequilibrium in order to understand past evolutionary and demographic events, to map genes that are associated with quantitative characters and inherited diseases, and to understand the joint evolution of linked sets of genes. This article introduces linkage disequilibrium, reviews the population genetic processes that affect it and describes some of its uses. At present, linkage disequilibrium is used much more extensively in the study of humans than in non-humans, but that is changing as technological advances make extensive genomic studies feasible in other species.

Linkage disequilibrium (LD) is one of those unfortunate terms that does not reveal its meaning. As every instructor of population genetics knows, the term is a barrier not an aid to understanding. LD means simply a nonrandom association of alleles at two or more loci, and detecting LD does not ensure either linkage or a lack of equilibrium. The term was first used in 1960 by Lewontin and Kojima[1] and it persists because LD was initially the concern of population geneticists who were not picky about terminology as long as the mathematical definition was clear. At first, there were few data with which to study LD, and its importance to evolutionary biology and human genetics was unrecognized outside of population genetics. However, interest in LD grew rapidly in the 1980s once the usefulness of LD for gene mapping became evident and large-scale surveys of closely linked loci became feasible. By then, the term was too well established to be replaced.

LD is of importance in evolutionary biology and human genetics because so many factors affect it and are affected by it. LD provides information about past events and it constrains the potential response to both natural and artificial selection. LD throughout the genome reflects the population history, the breeding system and the pattern of geographic subdivision, whereas LD in each genomic region reflects the history of natural selection, gene conversion, mutation and other forces that cause gene-frequency evolution. How these factors affect LD between a particular pair of loci or in a genomic region depends on local recombination rates. The population genetics theory of LD is well developed and is being widely used to provide insight into evolutionary history and as the basis for mapping genes in humans and in other species.

In this article, I will review the definitions of LD and the problems with assessing it, then outline the basic population genetics of LD that tells us how natural selection, genetic drift, recombination and mutation all affect levels of LD, and finally discuss some recent applications of LD to mapping genes, inferring the intensity of selection in the genome and estimating allele age.

Definitions

One pair of loci. LD between alleles at two loci has been defined in many ways (BOX 1), but all definitions depend on the quantity:

$$D_{AB} = p_{AB} - p_A p_B \tag{1}$$

which is the difference between the frequency of gametes carrying the pair of alleles A and B at two loci (p_{AB}) and the product of the frequencies of those alleles (p_A and p_B). Originally, the definition was in terms of gamete frequencies because that allows for the possibility that the

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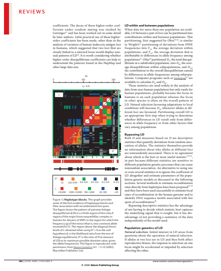

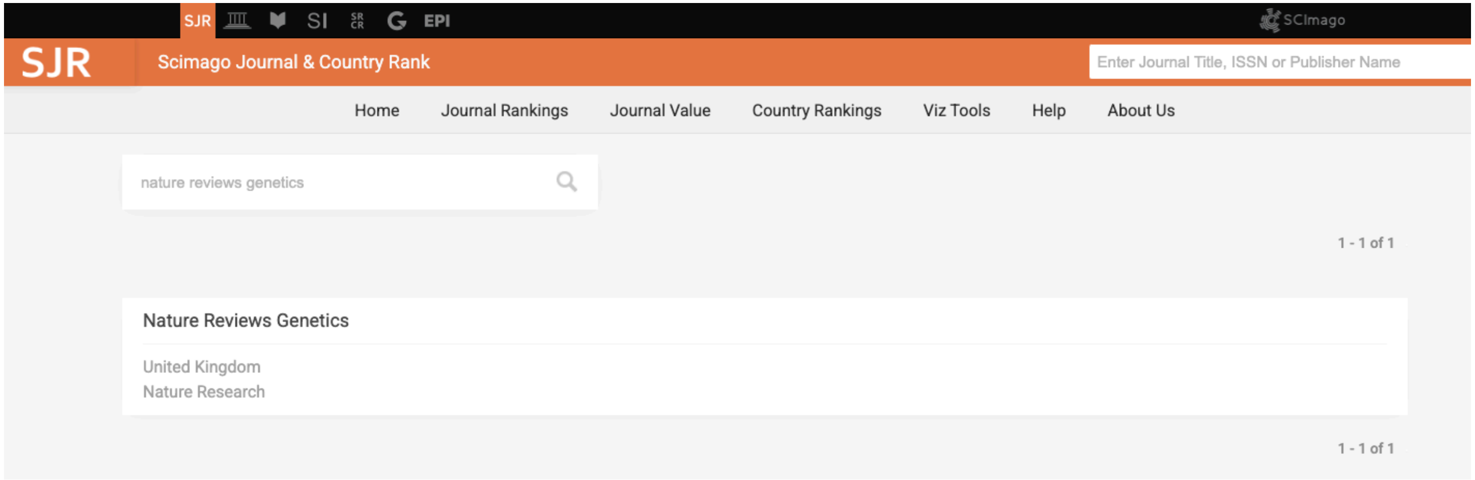

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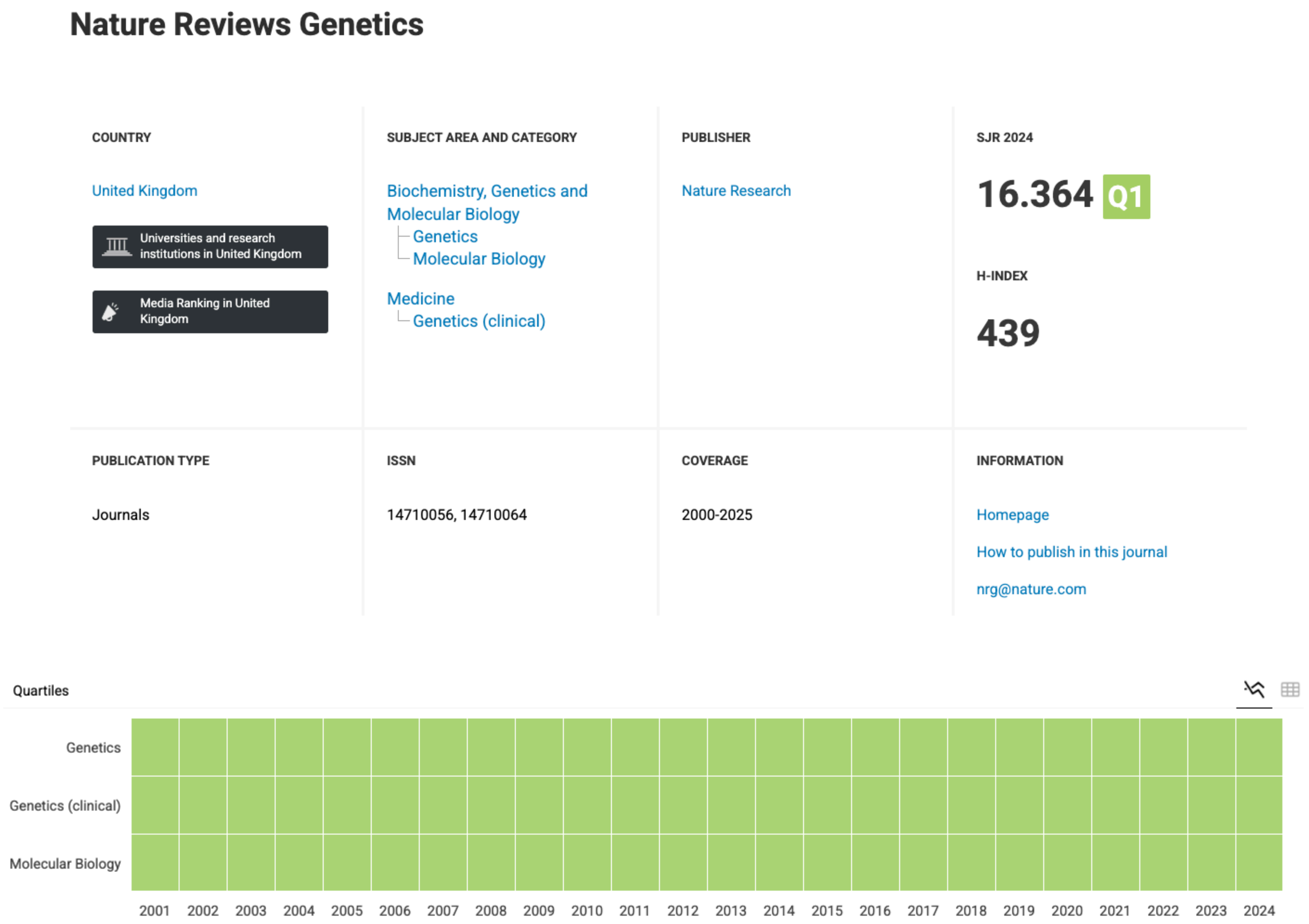

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RESEARCH ARTICLE

ANIMAL GENETICS WILEY

A *de novo* 2.3 kb structural variant in *MITF* explains a novel splashed white phenotype in a Thoroughbred family

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[3]Cell Physiology and Molecular Biophysics, School of Medicine, Texas Tech University Health Sciences Center, Lubbock, Texas, USA

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Abstract

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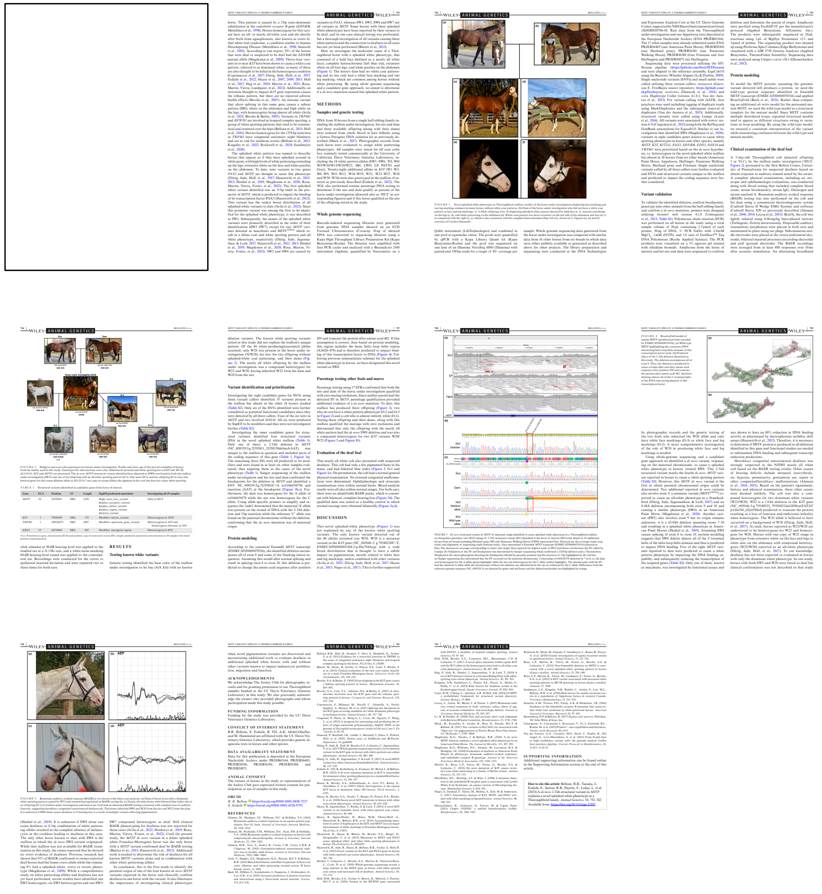
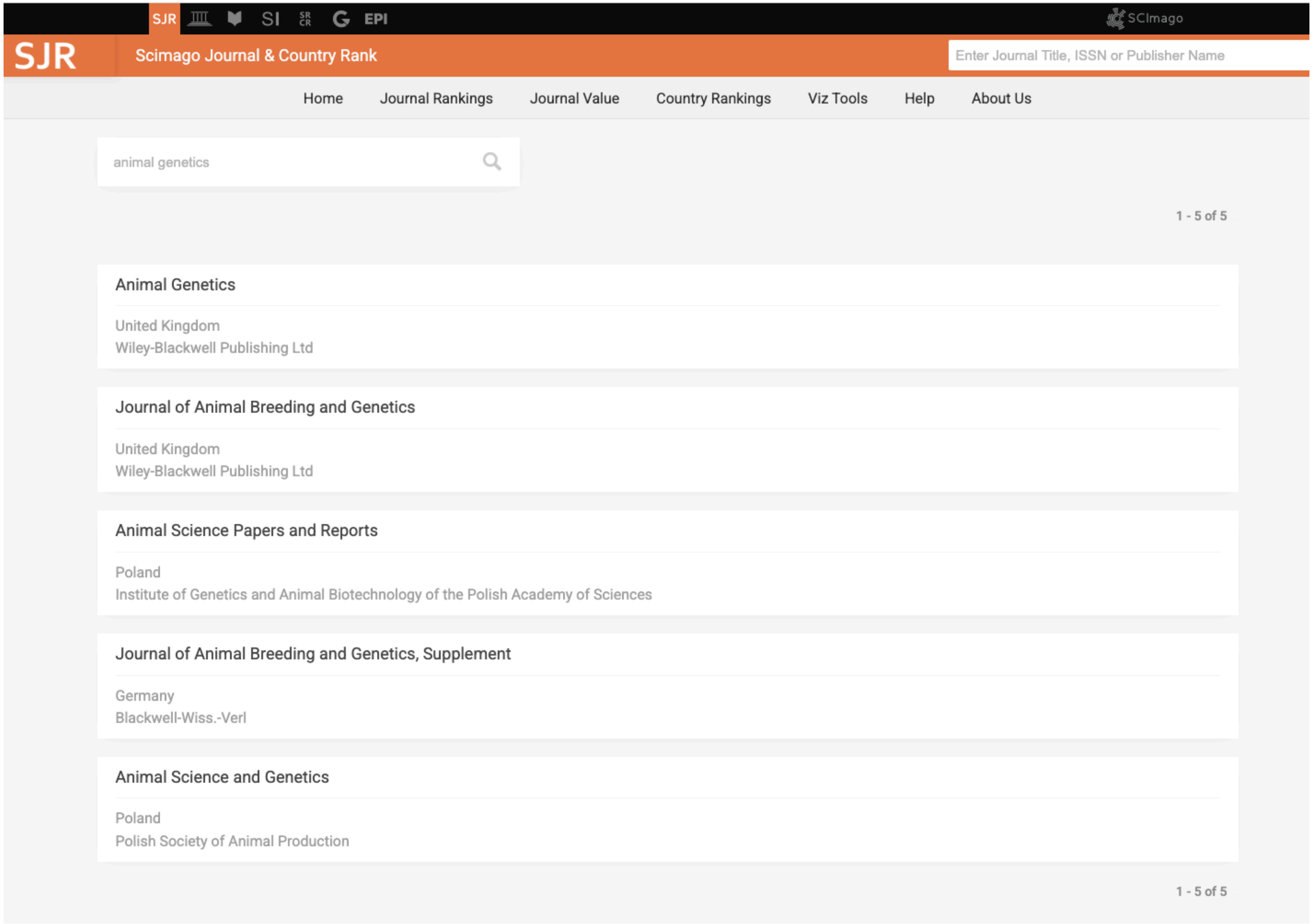
KEYWORDS
coat color, deafness, horse, pigmentation, white patterning

INTRODUCTION

Variation in white patterning has long fascinated animal enthusiasts and researchers alike. Moreover, some of these variations have a clinical connection and have similar counterparts in humans or other species. In horses, several different types of white patterning phenotypes have been described including frame overo, tobiano, roan, leopard complex spotting, dominant white, sabino and splashed white (Sponenberg & Bellone, 2017). The molecular bases of many of these have been determined, with the first to be unraveled being the frame overo pattern in which white patterning is prevalent on the ventral aspects of the horse with the pigment 'framing' the





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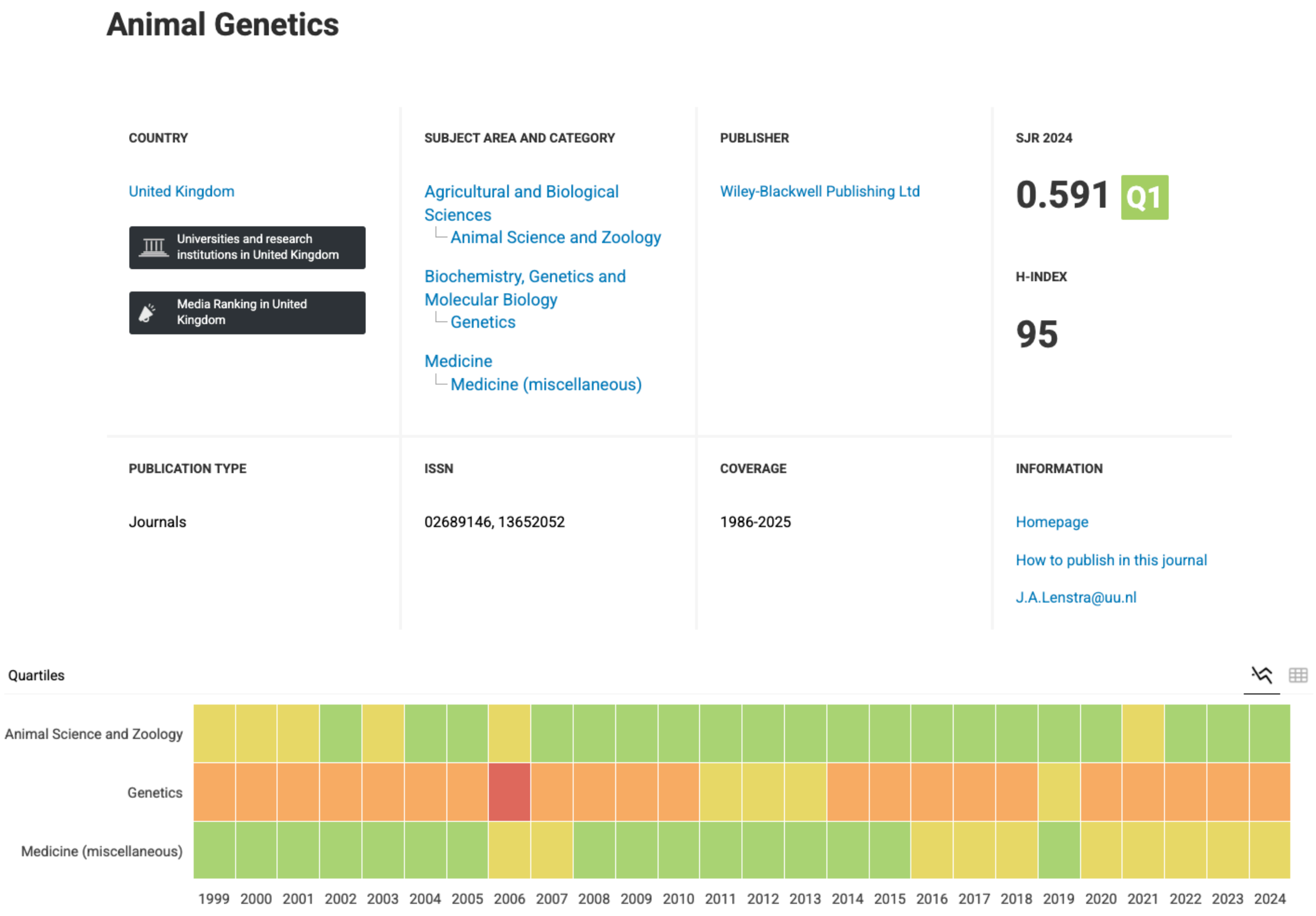

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Article analytics and metrics

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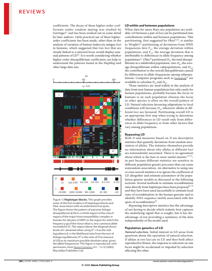

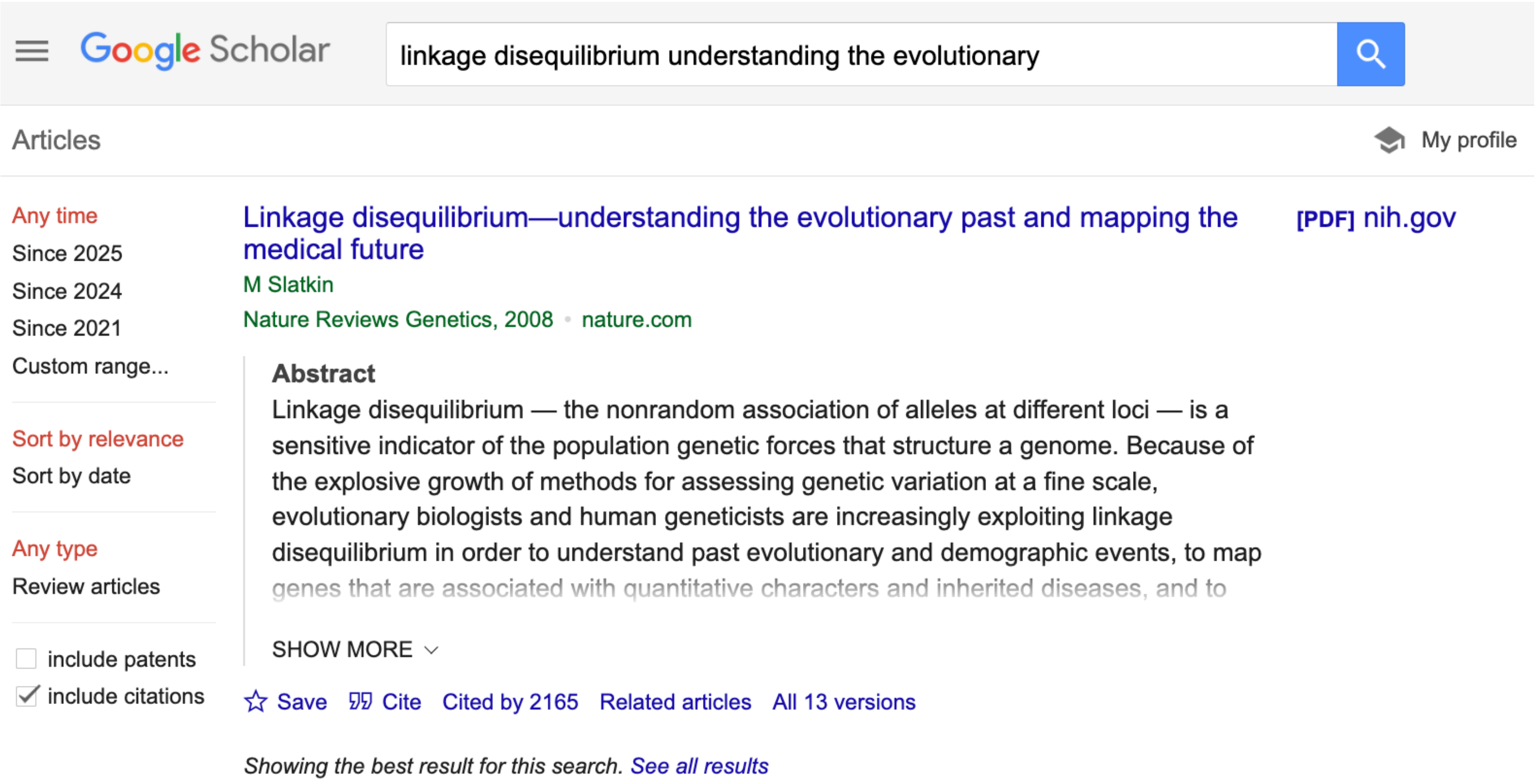

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Linkage disequilibrium—understanding the evolutionary past and mapping the medical future [PDF] nih.gov

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Montgomery Slatkin

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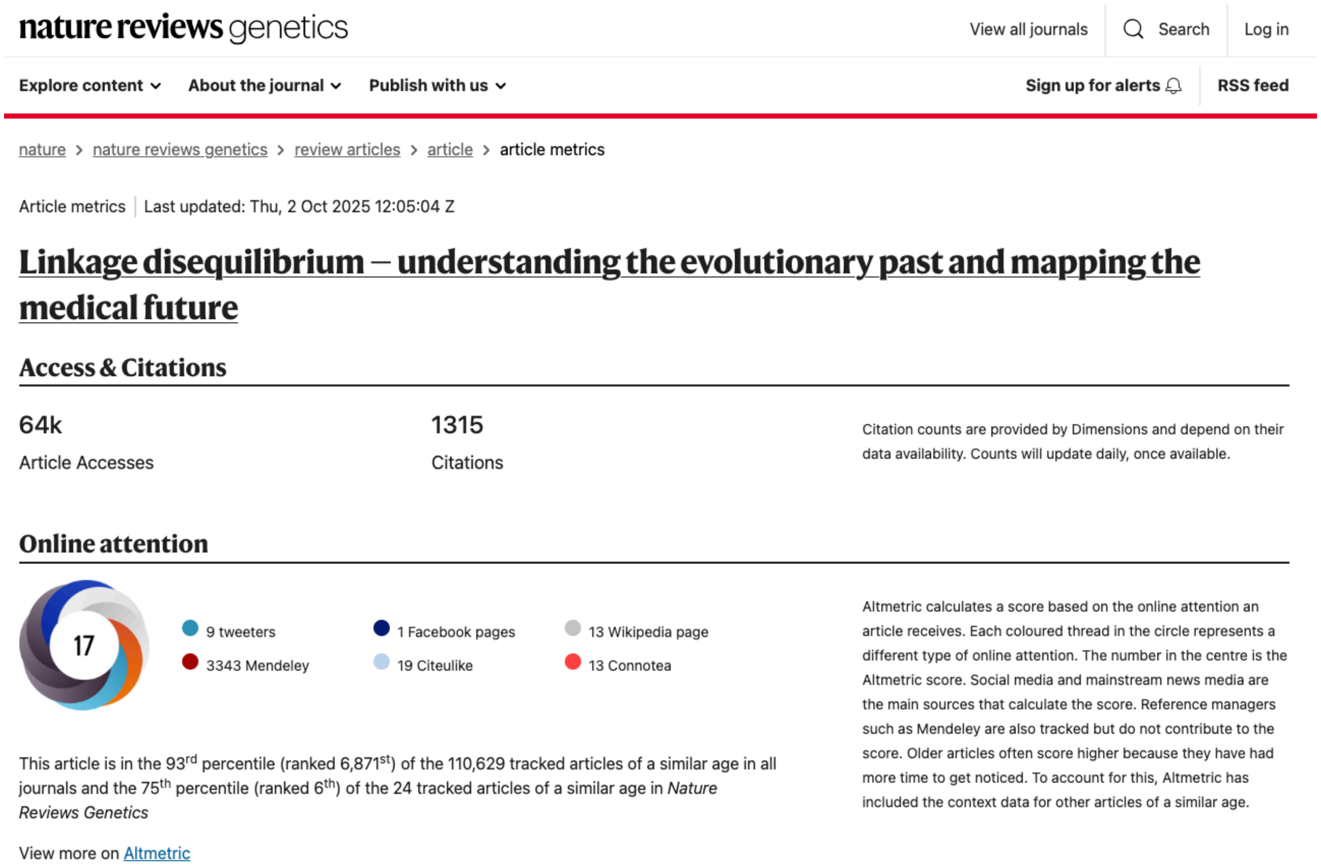

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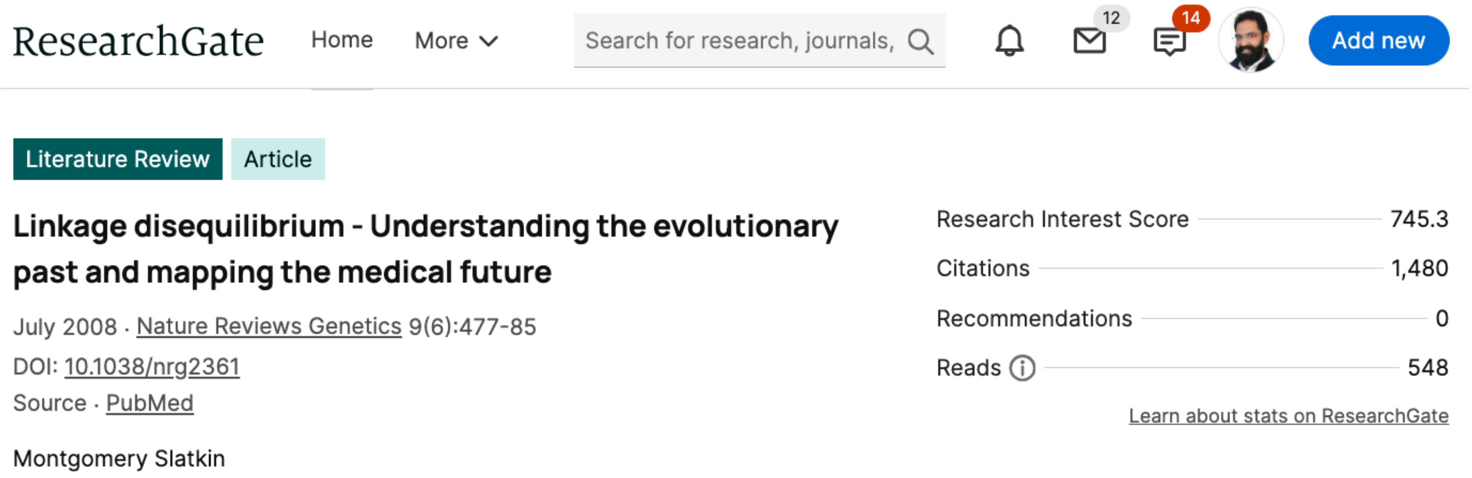

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RESEARCH ARTICLE

ANIMAL GENETICS WILEY

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Funding information
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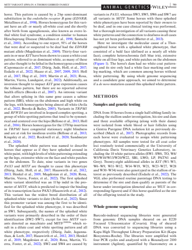
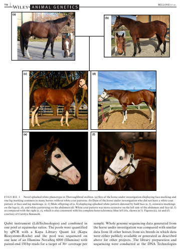

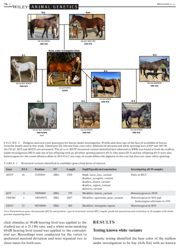

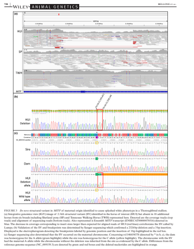
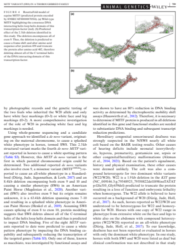
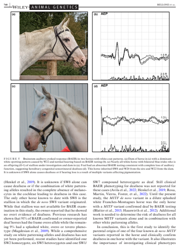
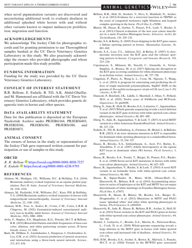


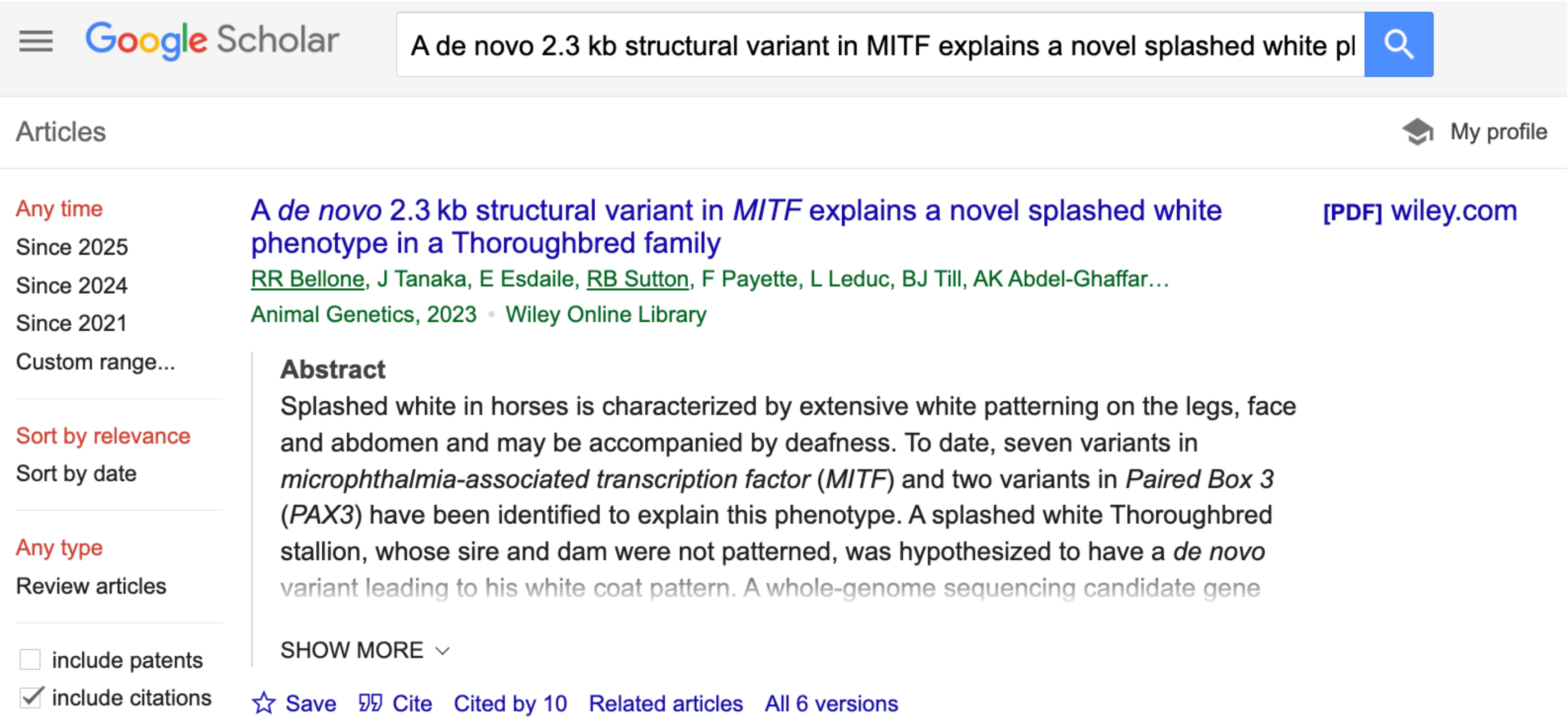

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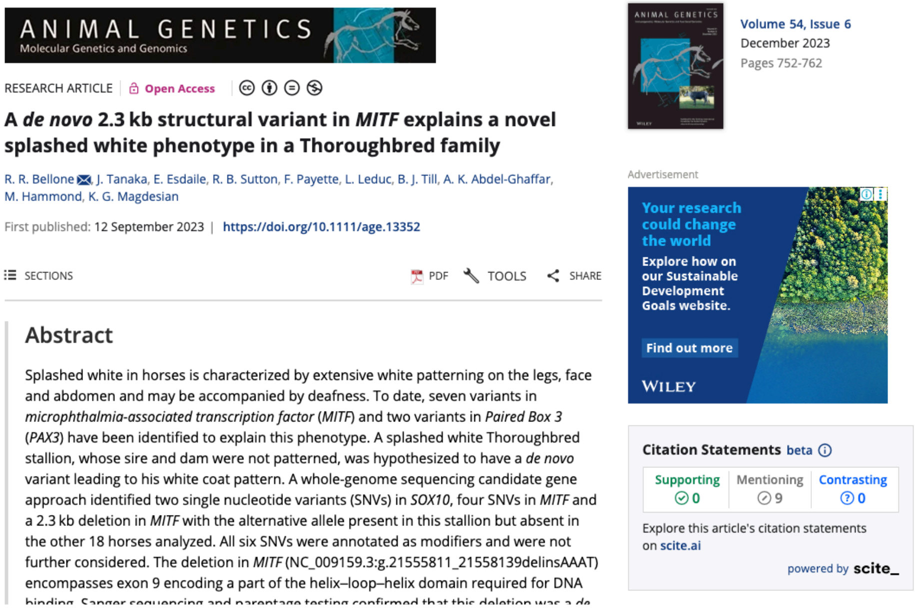

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A *de novo* 2.3 kb structural variant in *MITF* explains a novel splashed white phenotype in a Thoroughbred family

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Abstract: Splashed white in horses is characterized by extensive white patterning on the legs, face and abdomen and may be accompanied by deafness. To date, seven variants in microphthalmia-associated transcription factor (MITF) and two variants in Paired Box 3 (PAX3) have been identified to explain this phenotype. A splashed white Thoroughbred stallion, whose sire and dam were not patterned, was hypothesized to have a de novo variant leading to his white coat pattern. A whole-genome sequencing candidate gene approach i... Show more

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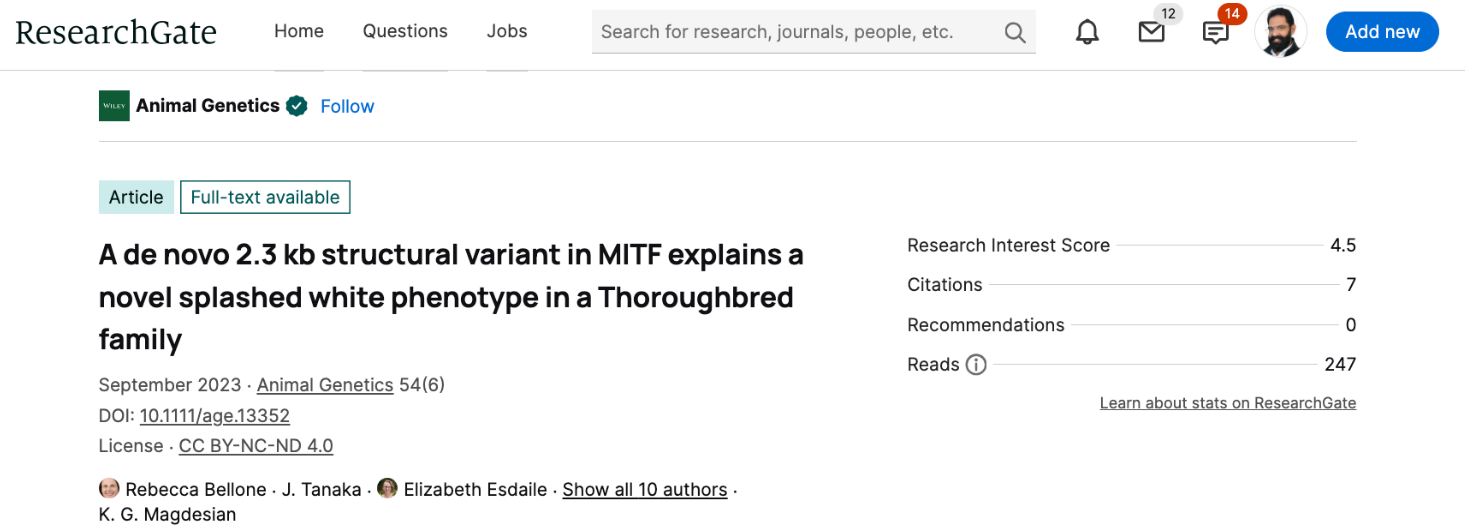

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September 2023 · Animal Genetics 54(6)
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FUNDAMENTAL CONCEPTS IN GENETICS

Linkage disequilibrium — understanding the evolutionary past and mapping the medical future

Montgomery Slatkin

Abstract | Linkage disequilibrium — the nonrandom association of alleles at different loci — is a sensitive indicator of the population genetic forces that structure a genome. Because of the explosive growth of methods for assessing genetic variation at a fine scale, evolutionary biologists and human geneticists are increasingly exploiting linkage disequilibrium in order to understand past evolutionary and demographic events, to map genes that are associated with quantitative characters and inherited diseases, and to understand the joint evolution of linked sets of genes. This article introduces linkage disequilibrium, reviews the population genetic processes that affect it and describes some of its uses. At present, linkage disequilibrium is used much more extensively in the study of humans than in non-humans, but that is changing as technological advances make extensive genomic studies feasible in other species.

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doi:10.1038/nrg2361

Linkage disequilibrium (LD) is one of those unfortunate terms that does not reveal its meaning. As every instructor of population genetics knows, the term is a barrier not an aid to understanding. LD means simply a nonrandom association of alleles at two or more loci, and detecting LD does not ensure either linkage or a lack of equilibrium. The term was first used in 1960 by Lewontin and Kojima[1] and it persists because LD was initially the concern of population geneticists who were not picky about terminology as long as the mathematical definition was clear. At first, there were few data with which to study LD, and its importance to evolutionary biology and human genetics was unrecognized outside of population genetics. However, interest in LD grew rapidly in the 1980s once the usefulness of LD for gene mapping became evident and large-scale surveys of closely linked loci became feasible. By then, the term was too well established to be replaced.

LD is of importance in evolutionary biology and human genetics because so many factors affect it and are affected by it. LD provides information about past events and it constrains the potential response to both natural and artificial selection. LD throughout the genome reflects the population history, the breeding system and the pattern of geographic subdivision, whereas LD in each genomic region reflects the history of natural selection, gene conversion, mutation and other forces that cause gene-frequency evolution. How these factors affect LD between a particular pair of loci or in a genomic region depends on local recombination rates. The population genetics theory of LD is well developed and is being widely used to provide insight into evolutionary history and as the basis for mapping genes in humans and in other species.

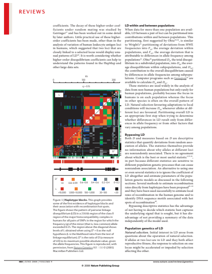
In this article, I will review the definitions of LD and the problems with assessing it, then outline the basic population genetics of LD that tells us how natural selection, genetic drift, recombination and mutation all affect levels of LD, and finally discuss some recent applications of LD to mapping genes, inferring the intensity of selection in the genome and estimating allele age.

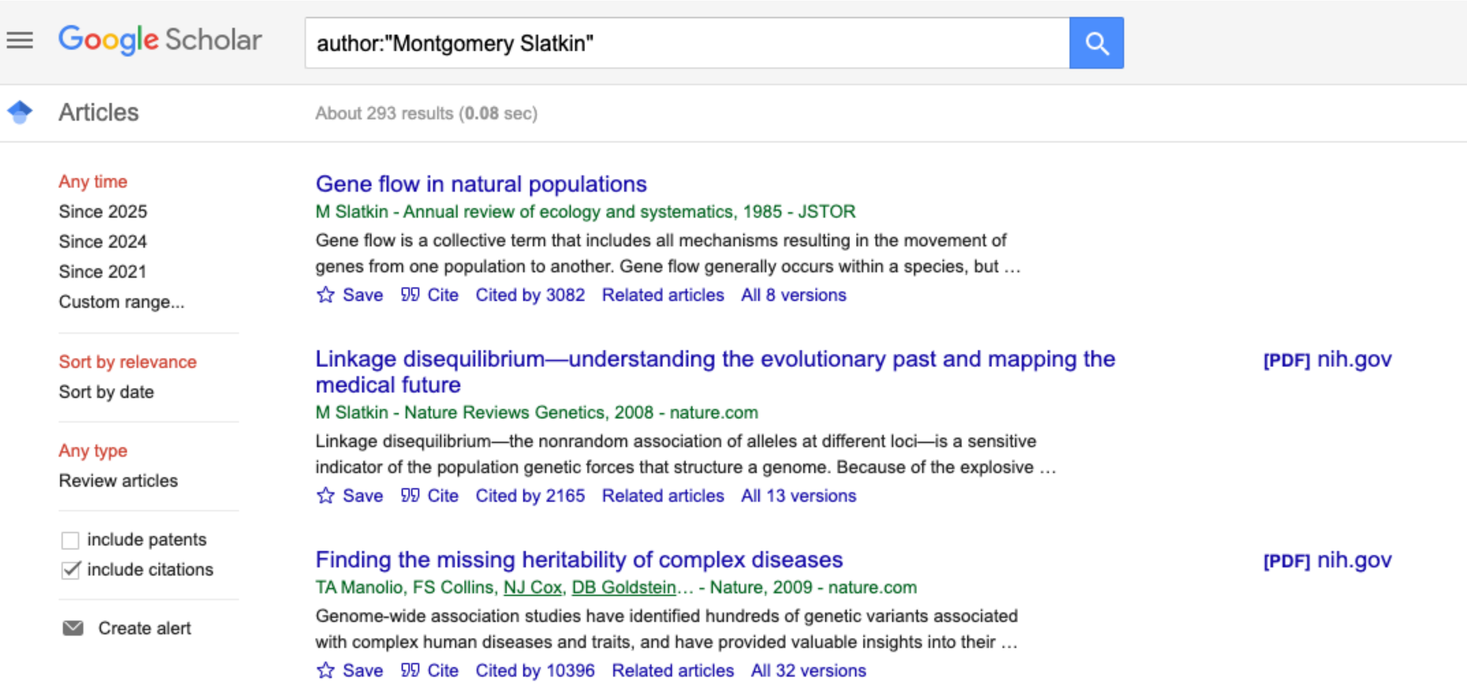
Definitions

One pair of loci. LD between alleles at two loci has been defined in many ways (BOX 1), but all definitions depend on the quantity:

$$D_{AB} = p_{AB} - p_A p_B \qquad (1)$$

which is the difference between the frequency of gametes carrying the pair of alleles A and B at two loci (p_{AB}) and the product of the frequencies of those alleles (p_A and p_B). Originally, the definition was in terms of gamete frequencies because that allows for the possibility that the





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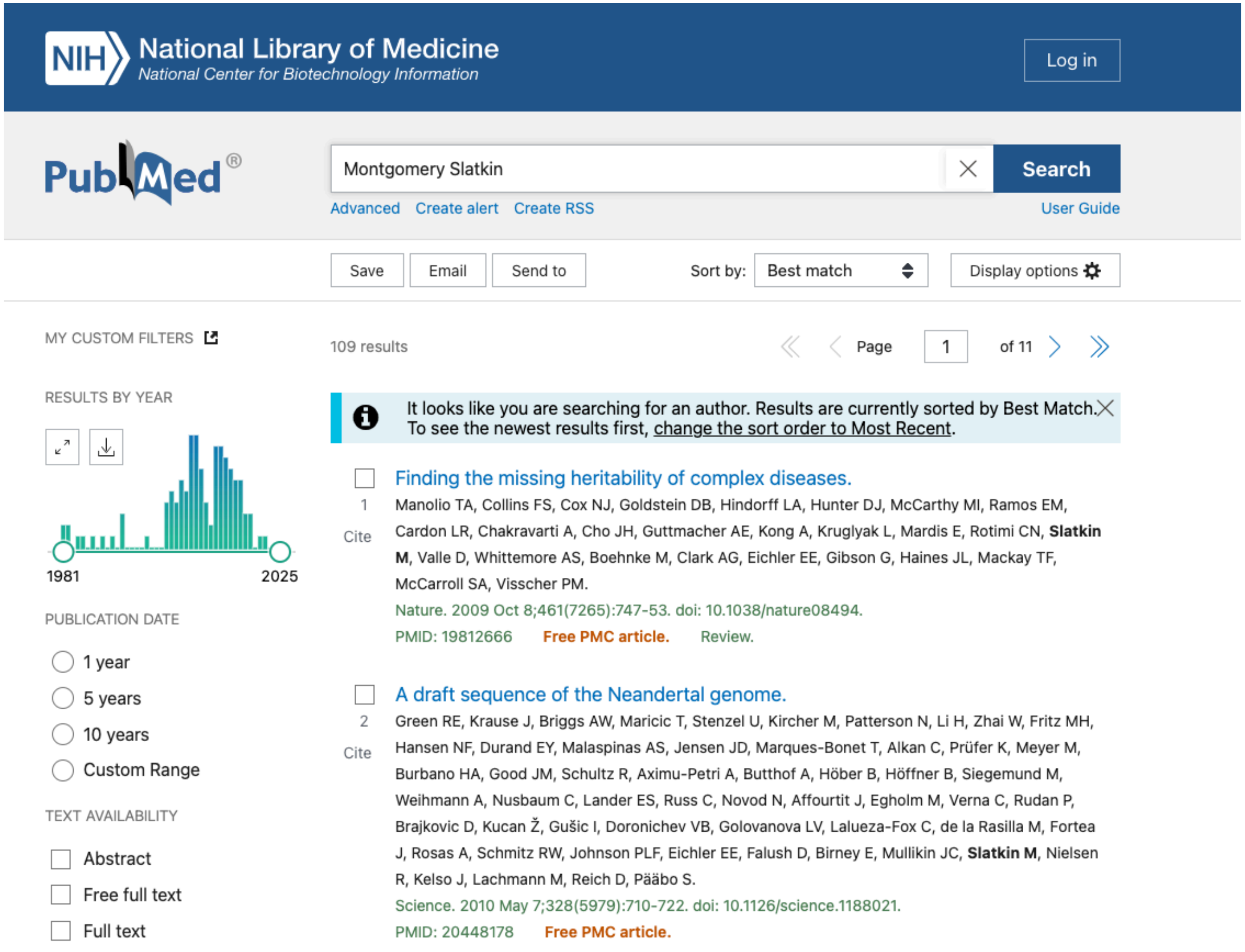

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Science. 2010 May 7;328(5979):710-722. doi: 10.1126/science.1188021.

PMID: 20448178 **Free PMC article.**

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RESEARCH ARTICLE

ANIMAL GENETICS WILEY

A *de novo* 2.3 kb structural variant in *MITF* explains a novel splashed white phenotype in a Thoroughbred family

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Email: rbellone@ucdavis.edu

Funding information
UC Davis Veterinary Genetics Laboratory

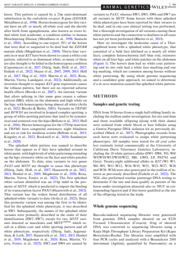
Abstract

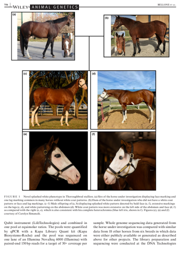
Splashed white in horses is characterized by extensive white patterning on the legs, face and abdomen and may be accompanied by deafness. To date, seven variants in *microphthalmia-associated transcription factor* (*MITF*) and two variants in *Paired Box 3* (*PAX3*) have been identified to explain this phenotype. A splashed white Thoroughbred stallion, whose sire and dam were not patterned, was hypothesized to have a *de novo* variant leading to his white coat pattern. A whole-genome sequencing candidate gene approach identified two single nucleotide variants (SNVs) in *SOX10*, four SNVs in *MITF* and a 2.3 kb deletion in *MITF* with the alternative allele present in this stallion but absent in the other 18 horses analyzed. All six SNVs were annotated as modifiers and were not further considered. The deletion in *MITF* (NC_009159.3:g.21555811_21558139delinsAAAT) encompasses exon 9 encoding a part of the helix–loop–helix domain required for DNA binding. Sanger sequencing and parentage testing confirmed that this deletion was a *de novo* mutation of maternal origin. Consistent with the published nomenclature, we denote this likely causal variant as SW8. Genotyping three of this stallion's offspring identified SW8 only in the nearly all-white foal that was confirmed deaf by brainstem auditory evoked response testing. This foal was also a compound heterozygote for dominant white variants (W20/W22), but to date, W variants alone have not been connected to deafness. SW8 marks the fourth de novo *MITF* variant in horses reported to cause white patterning. The link between deafness and all *MITF* variants with and without other variants impacting melanocyte development and function needs to be further explored.

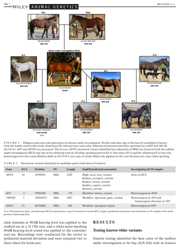
KEYWORDS
coat color, deafness, horse, pigmentation, white patterning

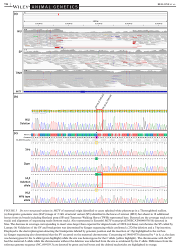
INTRODUCTION

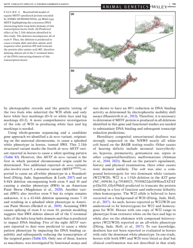
Variation in white patterning has long fascinated animal enthusiasts and researchers alike. Moreover, some of these variations have a clinical connection and have similar counterparts in humans or other species. In horses, several different types of white patterning phenotypes have been described including frame overo, tobiano, roan, leopard complex spotting, dominant white, sabino and splashed white (Sponenberg & Bellone, 2017). The molecular bases of many of these have been determined, with the first to be unraveled being the frame overo pattern in which white patterning is prevalent on the ventral aspects of the horse with the pigment 'framing' the

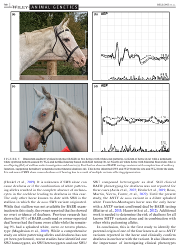


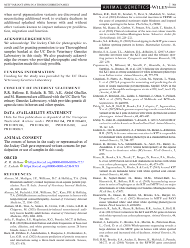


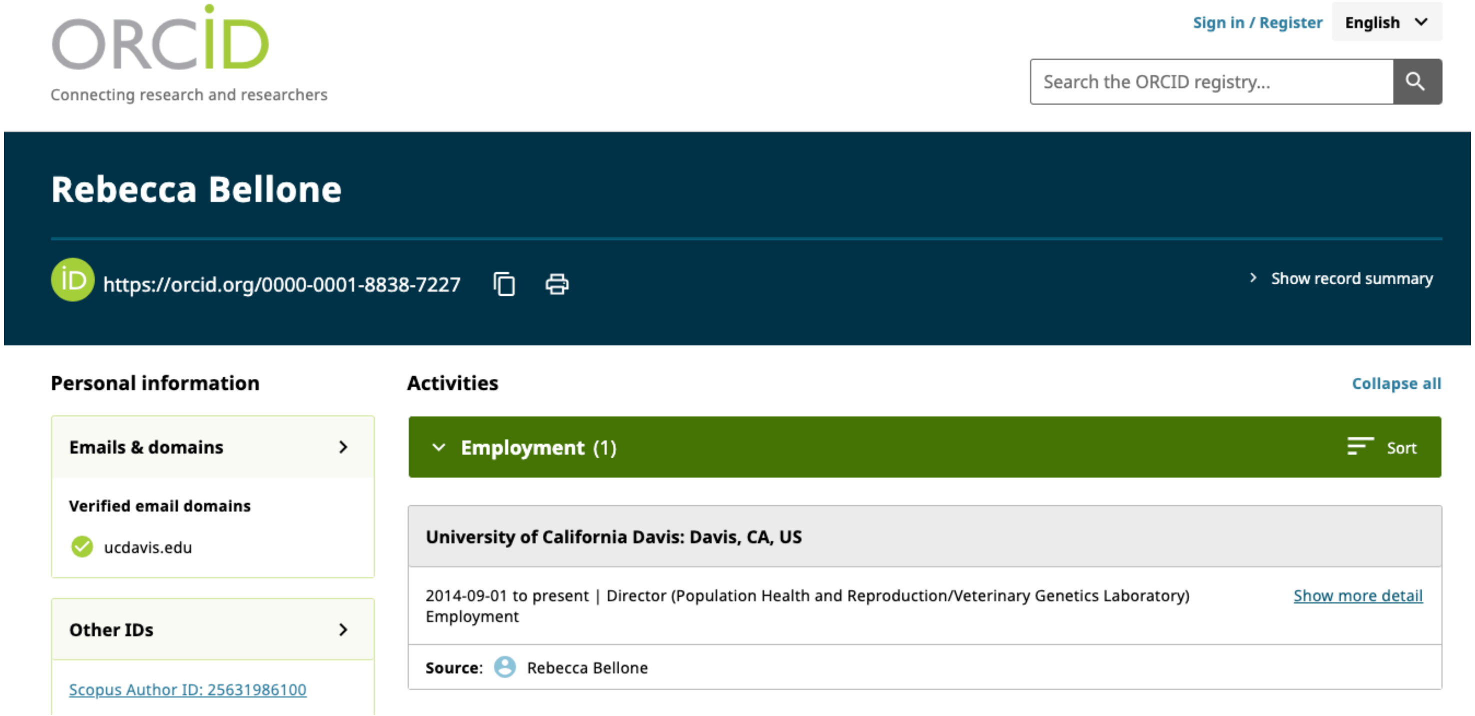












Rebecca Bellone

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Emails & domains

Verified email domains

✓ ucdavis.edu

Other IDs

Scopus Author ID: 25631986100

Activities

Employment (1)

University of California Davis: Davis, CA, US

2014-09-01 to present | Director (Population Health and Reproduction/Veterinary Genetics Laboratory)
Employment

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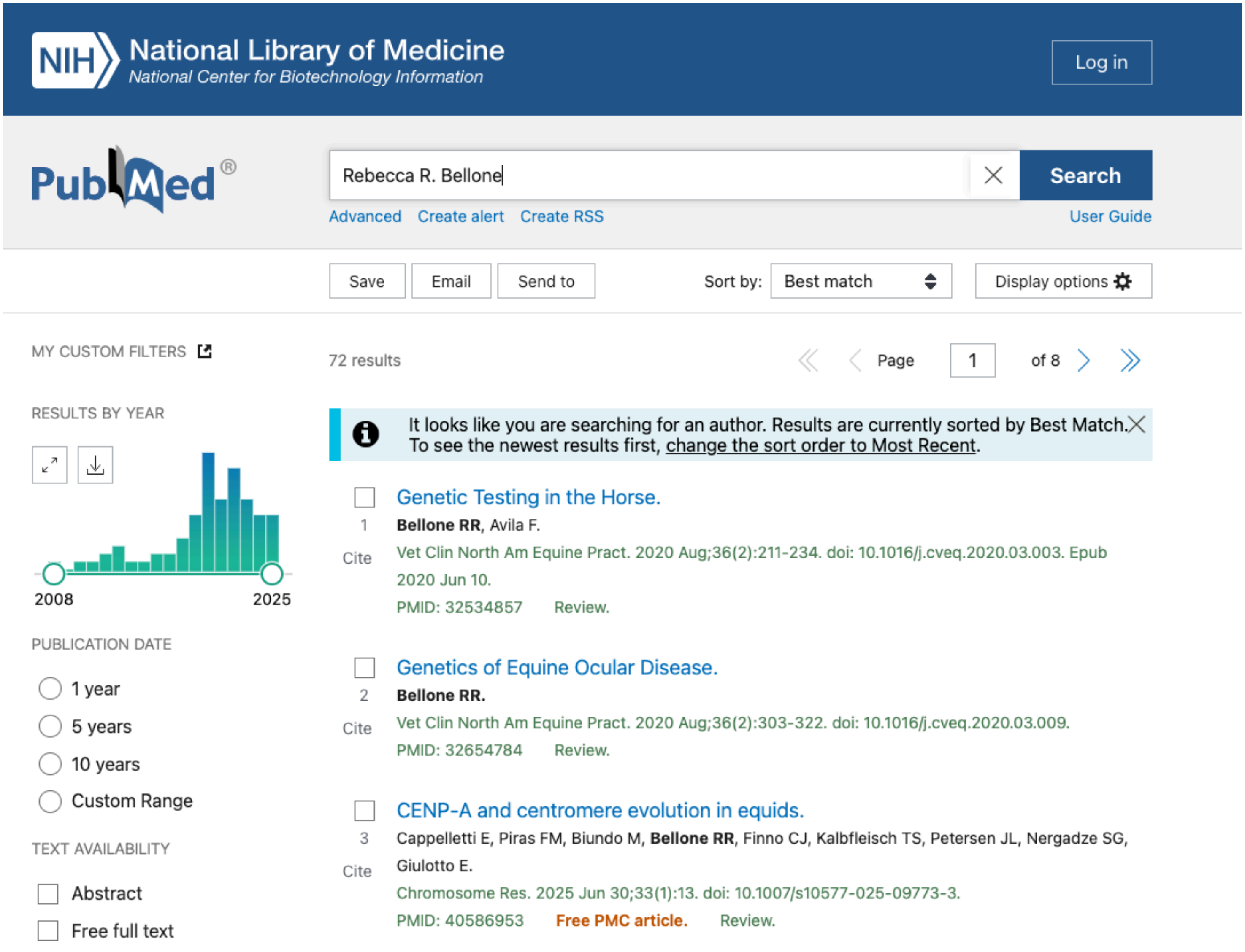

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Bellone RR, Avila F.

Vet Clin North Am Equine Pract. 2020 Aug;36(2):211-234. doi: 10.1016/j.cveq.2020.03.003. Epub 2020 Jun 10.

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Chromosome Res. 2025 Jun 30;33(1):13. doi: 10.1007/s10577-025-09773-3.

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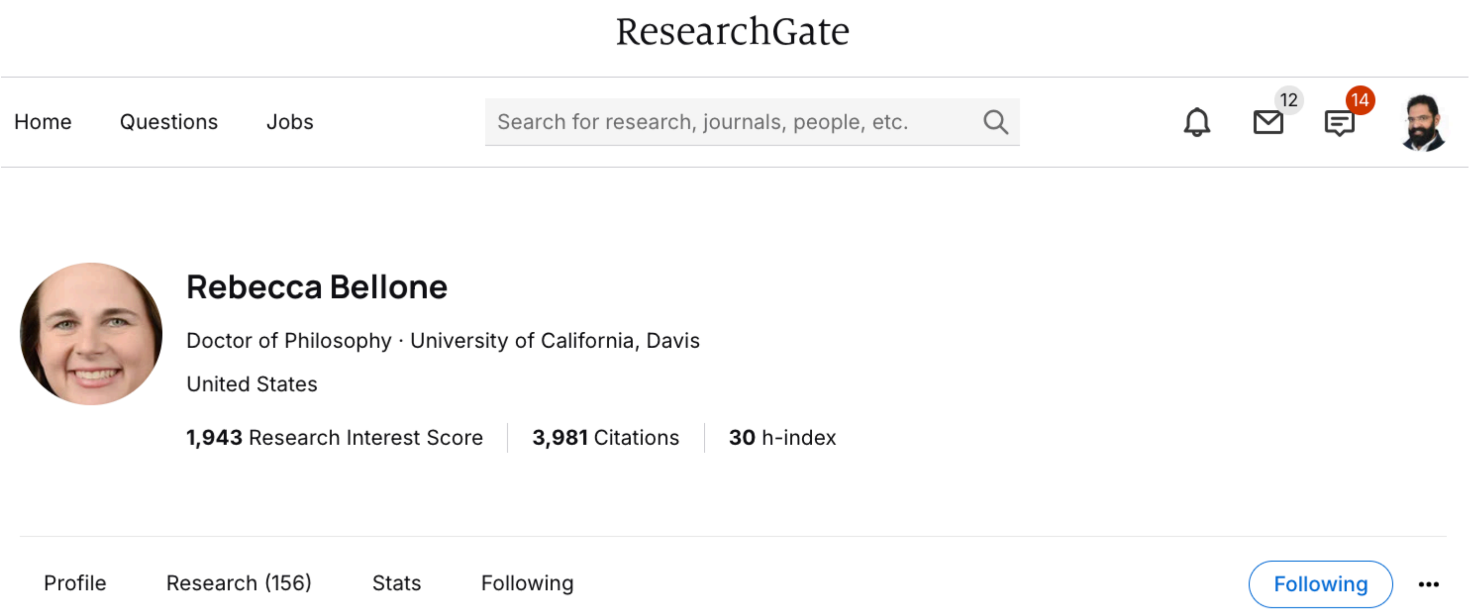

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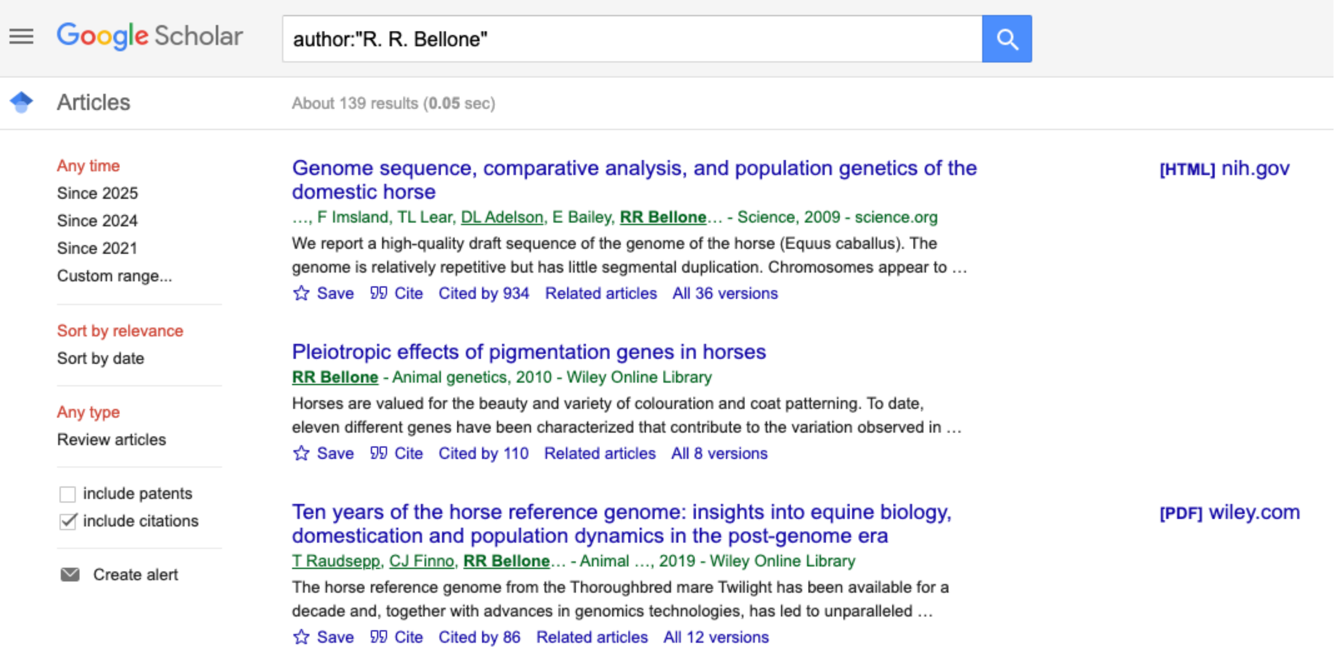

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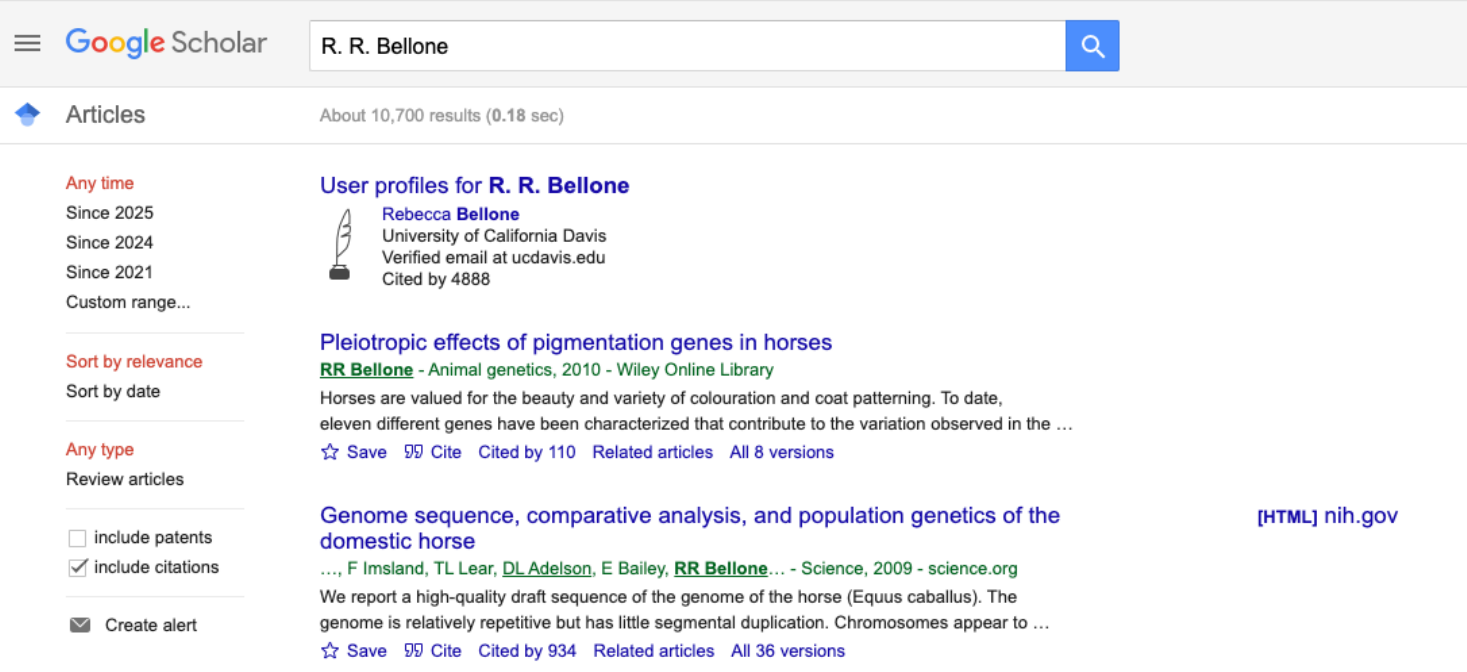

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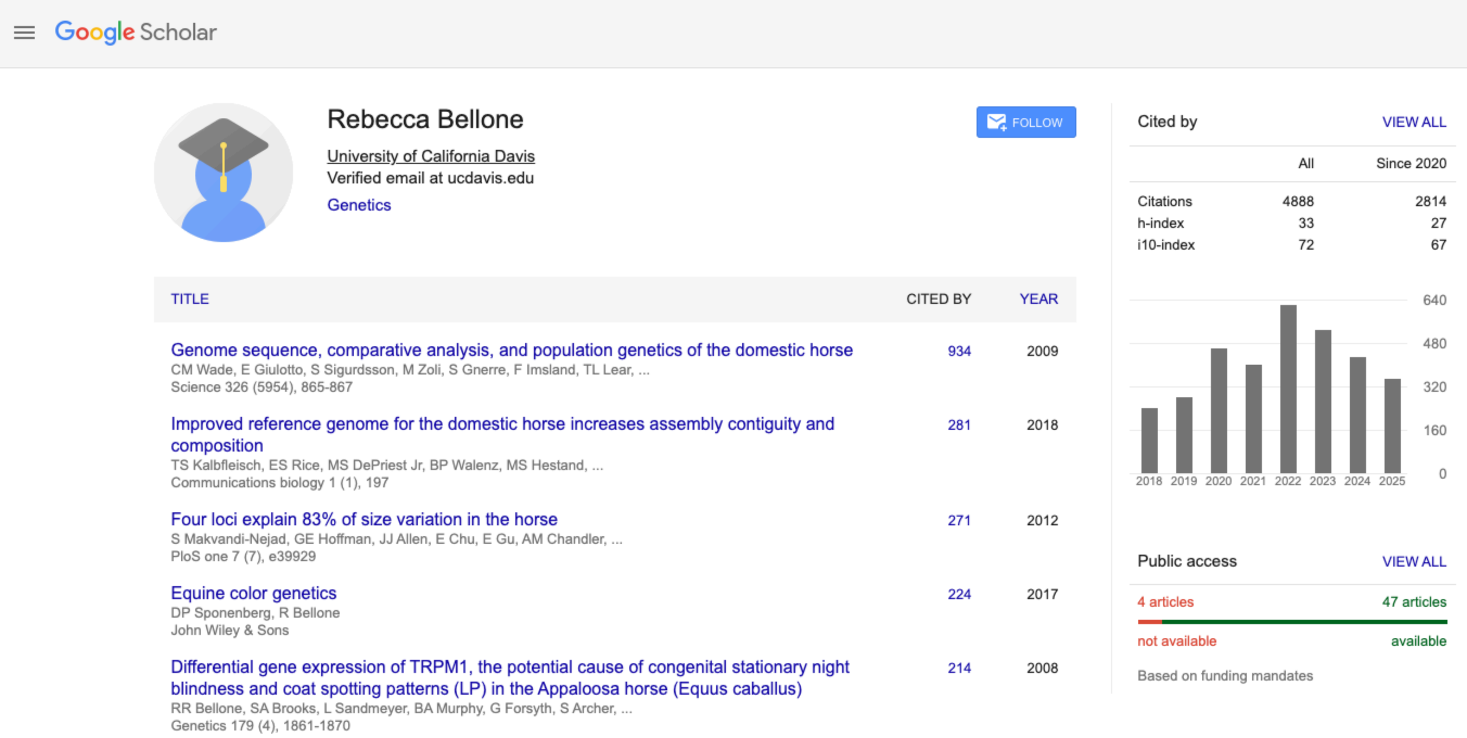

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Genome sequence, comparative analysis, and population genetics of the domestic horse CM Wade, E Giulotto, S Sigurdsson, M Zoli, S Gnerre, F Imsland, TL Lear, ... Science 326 (5954), 865-867	934	2009
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Student name: Mohammad Al Mutairi
Student ID: 222122341

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a. Publication

Ezo Emako, Seid Nuru, Mesfin Menza,
The effect of foreign direct investment on economic growth in developing countries,
Transnational Corporations Review,
Volume 14, Issue 4,
2022,

b. Paper metrics [2]

Citations	Year of publication
1	2022

c. Author metrics

Author	H. index	Number of publications	Number of citations
Emako, Ezo [3]	4	7	22
Nuru, Seid [4]	4	7	22
Menza, Mesfin [5]	4	8	35

a. Journal metrics [6] [7]

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Elsevier	22	Multidiscipline	Yes

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[1] https://www.sciencedirect.com/science/article/pii/S1925209924003218
[2] https://plu.mx/plum/a/?doi=10.1080/19186444.2022.2146967&theme=plum-sciencedirect-theme&hideUsage=true
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2. Review Article: Financial development and the growth effect of foreign direct investment: Does one size fit all? [8]

d. Publication

Michael J. Osei, Jaebeom Kim,
Financial development and the growth effect of foreign direct investment: Does one size fit all?,
International Economics,
Volume 173,
2023,

e. Paper metrics [9]

Citations	Year of publication
12	**2023**

f. Author metrics

Author	H. index	Number of publications	Number of citations
Osei, Michael J. [10]	**4**	**5**	**151**
Kim, Jaebeom [11]	**9**	**27**	**452**

b. Journal metrics [12] [13]

Publisher	H. index	Discipline	Open access
Elsevier	**22**	**Multidiscipline**	**Yes**

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[9] https://plu.mx/plum/a/?doi=10.1016/j.inteco.2023.01.001&theme=plum-sciencedirect-theme&hideUsage=true
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[13] https://scholar.google.com/citations?hl=en&view_op=search_venues&vq=Elsevier&btnG=

3. Short communication: FDI and economic growth: New evidence on the role of financial markets [14]

g. Publication

W.N.W. Azman-Saini, Siong Hook Law, Abd Halim Ahmad,
FDI and economic growth: New evidence on the role of financial markets,
Economics Letters,
Volume 107, Issue 2,
2010,

h. Paper metrics [15]

Citations	Year of publication
232	**2010**

i. Author metrics

Author	H. index	Number of publications	Number of citations
Azman-Saini, W. N.W. [16]	**20**	**86**	**1,880**
Law, Siong Hook [17]	**38**	**159**	**5,818**
Ahmad, Abd Halim [18]	**12**	**30**	**807**

c. Journal metrics [19] [20]

Publisher	H. index	Discipline	Open access
Elsevier	**22**	**Multidiscipline**	**Yes**

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