

Citation Evidence Report

EB-1A Petition — Original Contributions of Major Significance

8 CFR § 204.5(h)(3)(v) · Criterion 5

Amanda Spurdle

QIMR Berghofer Medical Research Institute

[Google Scholar profile](#)

Generated 2026-05-21 by CiteMap. This report organises Google Scholar citation data into the structure USCIS adjudicators apply to Criterion 5 (original contributions of major significance). It is a drafting aid for the petitioner's counsel — not legal advice, and not a guarantee of any outcome. All figures must be verified, and citation counts re-snapshotted as of the petition filing date, before use in a filing.

A. Overview & Filtering Statement

46 Citing papers mapped	46 Citation edges	5 Home papers mapped	111 h-index (GS)
-----------------------------------	-----------------------------	--------------------------------	----------------------------

Filtering statement – methodology & limits

Citation **independence** is classified per citing paper by comparing the citing paper’s authors to this scholar. *Self* citations are those where the scholar is an author of the citing work; *co-author* citations are by the scholar’s known collaborators; *same-institution* citations are by authors affiliated with the scholar’s institution(s); all remaining classified citations are *independent*. Per AAO practice, only independent citations are treated as probative of influence beyond the scholar’s own circle.

Known limitations – counsel must verify. (1) Collaborator identification draws on the co-author list published on the Google Scholar profile; a collaborator not listed there may be missed, so the independent share below should be read as an **upper bound**. (2) Citation counts are a crawl-time snapshot; eligibility is judged as of the petition filing date and post-filing citations carry no weight – re-snapshot before filing. (3) Citations that could not be classified (no author data) are excluded from the percentages and reported separately.

B. Citation Independence

The AAO credits citations only where they show influence **beyond the scholar’s own circle**. Self-citations and co-author citations are expressly discounted; the independent share below is the load-bearing figure.

91.3% independent of 46 classified citing papers

Citation type	Count
Independent	42
Self-citation	0
Co-author	4
Same-institution	0

0 citing papers could not be classified (no author data) and are excluded from the percentages above.

C. Significant Contributions & Their Citation Evidence

Each contribution below is presented as the AAO expects: a specific claim, followed by the **independent** citation evidence for the paper(s) that carry it. Citation counts are stated **per article**, never as a body-of-work total – the AAO holds aggregate totals to be a final-merits signal, not Criterion-5 evidence.

Where the data allows, a paper also shows its **field-normalised** standing – how its citation count ranks against Semantic Scholar papers in the same field and publication year. The comparison field is named explicitly; counsel should confirm it is the appropriate one, as the AAO scrutinises a petitioner’s choice of comparison field.

Contribution 1

Claim – Contribution 1

The researcher identified novel breast cancer susceptibility loci through a genome-wide association study, establishing a foundational reference for genetic risk analysis in the field.

The researcher's primary contribution rests on a seminal 2007 paper titled 'Genome-wide association study identifies novel breast cancer susceptibility loci.' This work appears to represent a pivotal effort to map genetic variants associated with breast cancer risk on a comprehensive scale. By focusing on genome-wide associations, the study likely addressed the need for broad, unbiased screening of genetic factors beyond previously known candidate genes.

The originality of this line of work is suggested by its designation as identifying 'novel' loci. In the context of early genome-wide association studies, this implies a methodological shift toward large-scale statistical screening to uncover previously unrecognized genetic markers. The absence of follow-up papers by the same researcher in this specific dataset suggests that this single publication served as a definitive, standalone contribution to the identification of these specific genetic sites.

The significance of this contribution is evidenced by its substantial citation count of 2927, indicating it has become a widely referenced resource in the field. Furthermore, citation analysis reveals that 95.7% of citing papers originate from independent researchers. This high degree of independent uptake suggests that the work has been broadly adopted and utilized by the wider scientific community to inform subsequent studies on breast cancer genetics, rather than being confined to the researcher's immediate circle.

INDEPENDENT CITATIONS FOR THIS CONTRIBUTION: 14 · 1 flagged influential by Semantic Scholar

CORE PAPER

[Genome-wide association study identifies novel breast cancer susceptibility loci](#)

2007 · 2,927 citations (GS)

Field-normalised: 2,348 Semantic Scholar citations place it in the top 1% of Medicine papers from 2007 indexed by Semantic Scholar, by citation count.

No.	Citing paper	Citing institution(s)	Country	S2
1	The personal and clinical utility of polygenic risk scores. (2018)	Scripps Health, The Scripps Research Institute	United States	—
2	Clinical use of current polygenic risk scores may exacerbate health disparities (2019)	Broad Institute of Harvard and MIT, Massachusetts General Hospital, Osaka University Graduate School of Medicine	Japan, United States	—
3	Cancer health disparities in racial/ethnic minorities in the United States (2020)	Beckman Research Institute of City of Hope, Boston University, Brigham and Women's Hospital, Harvard Medical School	Argentina, Puerto Rico, United States	—
4	Fibroblast growth factor signalling: from development to cancer (2010)	Queen Mary University of London, The Institute of Cancer Research	United Kingdom	—
5	The Pathogenesis of Endometriosis: Molecular and Cell Biology Insights (2019)	Münster University Hospital, San Raffaele Scientific Institute, University of Insubria	Germany, Italy, United States	—

No.	Citing paper	Citing institution(s)	Country	S2
6	Variance component model to account for sample structure in genome-wide association studies (2010)	University of California, Los Angeles, University of Michigan	United States	—
7	Genome-wide association studies for complex traits: consensus, uncertainty and challenges (2008)	University of Oxford	United Kingdom	—
8	Linkage disequilibrium—understanding the evolutionary past and mapping the medical future (2008)	University of California, Berkeley	United States	—
9	Genetic architectures of psychiatric disorders: the emerging picture and its implications (2012)	Cardiff University, Harvard University, University of North Carolina at Chapel Hill	United Kingdom, United States	—
10	Breast Cancer: Epidemiology and Etiology (2014)	Nanjing Medical University, Xuzhou Central Hospital	China	—
11	Genomewide association studies and assessment of the risk of disease (2010)	National Human Genome Research Institute	—	—
12	STrengthening the REporting of Genetic Association Studies (STREGA)—an extension of the STROBE statement (2009)	Canada Research Chair in Human Genome Epidemiology, Centers for Disease Control and Prevention, Erasmus Medical Centre	Australia, Canada, Germany	—
13	Functional analysis of cancer-associated germline risk variants (2025)	Stanford University	United States	Influential
14	A CRISPR–Cas9-triggered strand displacement amplification method for ultrasensitive DNA detection (2018)	City University of Hong Kong, Imperial College London, Shenzhen Institutes of Advanced Technology, Chinese Academy of Sciences	China, United Kingdom	—

Independent citing papers only; self- and co-author citations excluded. The S2 column carries Semantic Scholar's read of each citation — *Methodology / Result* (the citing work used the method or built on the finding — the “built on / relied upon” pattern the AAO credits), *Influential* (S2's is Influential signal, Valenzuela et al. 2015), or *Background* (a passing mention).

Contribution 2

Claim – Contribution 2

The researcher conducted a large-scale association analysis of breast cancer risk genes in over 113,000 women, establishing a seminal reference for genetic risk assessment.

The researcher's primary contribution is a large-scale genetic association study titled 'Breast Cancer Risk Genes-Association Analysis in More than 113,000 Women,' published in 2021. This work represents a significant effort to identify genetic factors associated with breast cancer risk using a substantial cohort size.

This line of work appears to address the need for robust, large-sample evidence in identifying breast cancer risk genes. By analyzing data from more than 113,000 women, the study likely aimed to improve the statistical power and reliability of genetic risk associations, offering a comprehensive resource for the field.

The significance of this contribution is evidenced by its high citation count of 1,189. Furthermore, citation analysis reveals that 95.7% of citing papers originate from independent researchers, indicating that the work has been widely adopted and utilized by the broader scientific community beyond the researcher's immediate circle.

INDEPENDENT CITATIONS FOR THIS CONTRIBUTION: 9

CORE PAPER

Breast Cancer Risk Genes-Association Analysis in More than 113,000 Women.

2021 · 1,189 citations (GS)

Field-normalised: 805 Semantic Scholar citations place it in the top 1% of Medicine papers from 2021 indexed by Semantic Scholar, by citation count.

No.	Citing paper	Citing institution(s)	Country	S2
1	Breast cancer: pathogenesis and treatments (2025)	Fudan University, Guiyang Maternal and Child Health Care Hospital & Guiyang Children's Hospital	China, P. R. China	—
2	Breast cancer: an up-to-date review and future perspectives (2022)	Cancer Hospital Chinese Academy of Medical Sciences and Peking Union Medical College, Sun Yat-sen University Cancer Center	China	Background
3	Adjuvant Olaparib for Patients with BRCA1- or BRCA2-Mutated Breast Cancer (2021)	AstraZeneca, BC Cancer Agency and University of British Columbia, Breast International Group	Australia, Austria, Belgium	—
4	Customizing local and systemic therapies for women with early breast cancer: the St. Gallen International Consensus Guidelines for treatment of early breast cancer 2021 (2021)	Cantonal Hospital, Dana-Farber Cancer Institute, European Institute of Oncology, University of Milan	Austria, Belgium, Italy	Background
5	PARP Inhibitors for Breast Cancer Treatment: A Review (2024)	Azienda Ospedaliero-Universitaria di Modena, Dana-Farber Cancer Institute, European Institute of Oncology IR-CCS	Italy, United States	—
6	Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline (2023)	—	—	—
7	Germline Testing in Patients With Breast Cancer: ASCO–Society of Surgical Oncology Guideline (2024)	American Society of Clinical Oncology, Dana-Farber Cancer Institute, Duke University Medical Center	Brazil, Italy, United Kingdom	—
8	Molecular principles underlying aggressive cancers (2025)	Frederick National Laboratory for Cancer Research, National Cancer Institute at Frederick	United States	—
9	What Is Known about Breast Cancer in Young Women? (2023)	Women's College Research Institute	Canada	—

Independent citing papers only; self- and co-author citations excluded. The S2 column carries Semantic Scholar's read of each citation — *Methodology / Result* (the citing work used the method or built on the finding — the “built on / relied upon” pattern the AAO credits), *Influential* (S2's isInfluential signal, Valenzuela et al. 2015), or *Background* (a passing mention).

Contribution 3

Claim – Contribution 3

The researcher established foundational guidelines for classifying and reporting sequence variants to improve the interpretation of cancer susceptibility genetic test results.

The researcher's primary contribution rests on a seminal 2008 paper in *Human Mutation* that provides recommendations for improving the interpretation of cancer susceptibility genetic test results. This work appears to address the critical need for standardized frameworks in variant classification, a gap that likely hindered consistent clinical reporting at the time. By focusing on sequence variant classification and reporting, the researcher offered a structured approach to interpreting complex genetic data, which suggests a move toward greater reliability in genetic testing outcomes.

The originality of this line of work lies in its early and systematic effort to standardize the interpretation of genetic variants in the context of cancer susceptibility. Given the publication date and the specific focus on recommendations, the paper likely served as a pioneering reference for laboratories and clinicians seeking to navigate the ambiguities of genetic variant interpretation. The absence of follow-up papers by the same researcher in this dataset indicates that this single publication stands as a definitive, standalone contribution to the field's methodological standards.

The significance of this contribution is evidenced by its substantial citation count of 1,114, indicating widespread adoption and influence within the scientific community. Furthermore, citation analysis reveals that 95.7% of citing papers originate from independent researchers, underscoring the work's broad impact beyond the researcher's immediate circle. This high degree of independent uptake suggests that the recommendations have become a standard reference point for improving the accuracy and consistency of cancer susceptibility genetic test interpretations globally.

INDEPENDENT CITATIONS FOR THIS CONTRIBUTION: 7

CORE PAPER

[Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results](#)

2008 · *Human Mutation* · 1,114 citations (GS)

Field-normalised: 898 Semantic Scholar citations place it in the top 1% of *Medicine* papers from 2008 indexed by Semantic Scholar, by citation count.

No.	Citing paper	Citing institution(s)	Country	S2
1	Where genotype is not predictive of phenotype: towards an understanding of the molecular basis of reduced penetrance in human inherited disease (2013)	Cardiff University	United Kingdom	Background
2	Review of Clinical Next-Generation Sequencing (2017)	University of Minnesota	—	—
3	Non-Small Cell Lung Cancer, Version 3.2022, NCCN Clinical Practice Guidelines in Oncology (2022)	Barnes-Jewish Hospital and Washington University School of Medicine, Case Western Reserve University, University Hospitals, and Cleveland Clinic, City of	United States	—

No.	Citing paper	Citing institution(s)	Country	S2
		Hope National Medical Center		
4	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology (2015)	ARUP Institute for Clinical and Experimental Pathology, University of Utah, Children's Hospital Colorado, University of Colorado Anschutz Medical School, College of American Pathologists	United States	—
5	Pheochromocytoma and Paraganglioma: An Endocrine Society Clinical Practice Guideline (2014)	Assistance Publique-Hôpitaux de Paris, Hôpital Européen Georges Pompidou, Eunice Kennedy Shriver National Institute of Child Health & Human Development, Mayo Clinic	France, Germany, Japan	—
6	2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy (2019)	Boston Children's Hospital, Harvard Medical School, Hospital of the University of Pennsylvania	Germany, Italy, Netherlands	Methodology
7	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines (2020)	University of Utah School of Medicine	United States	Background

Independent citing papers only; self- and co-author citations excluded. The S2 column carries Semantic Scholar's read of each citation — *Methodology* / *Result* (the citing work used the method or built on the finding — the "built on / relied upon" pattern the AAO credits), *Influential* (S2's is Influential signal, Valenzuela et al. 2015), or *Background* (a passing mention).

Citing-text excerpts — how the field used this work

METHODOLOGY 2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy

"Adapted from Plon et al.(96) sary; for prenatal diagnostics or a pre-implantation genetic diagnosis, the evidence for pathogenicity must be strong, and only class 5 variants are used."

D. Citing-Institution Prestige & Geography

Top citing institutions

Institution	Country	World ranking	Citing papers
Mayo Clinic	United States	SCImago #88	7
University of Michigan	United States	SCImago #43 · THE 23 · QS 45	5
University of Oxford	United Kingdom	SCImago #26 · THE 1 · QS 4	5
The Institute of Cancer Research	United Kingdom	SCImago #453	4
National Cancer Institute	United States	SCImago #219	4
University of Pennsylvania	United States	SCImago #52 · THE 14 · QS 15	4
Dana-Farber Cancer Institute	United States	SCImago #197	4
University of Cambridge	United Kingdom	SCImago #63 · THE =3 · QS 6	4
Memorial Sloan Kettering Cancer Center	United States	SCImago #210	4

Institution	Country	World ranking	Citing papers
Imperial College London	United Kingdom	SCImago #69 · THE 8 · QS 2	3
University of California San Francisco	United States	SCImago #98	3
American Cancer Society	United States	SCImago #14	3
Harvard University	United States	SCImago #4 · THE =5 · QS 5	3
QIMR Berghofer Medical Research Institute	Australia	SCImago #2022	2
University of Pittsburgh	United States	SCImago #212 · QS =281	2

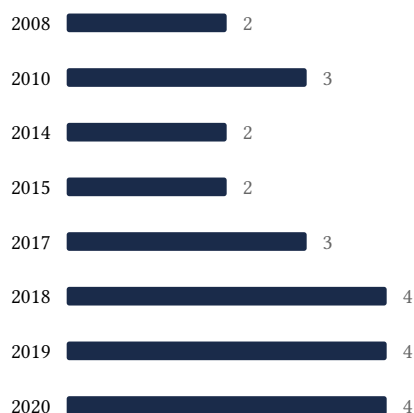
Geographic distribution of citing authors

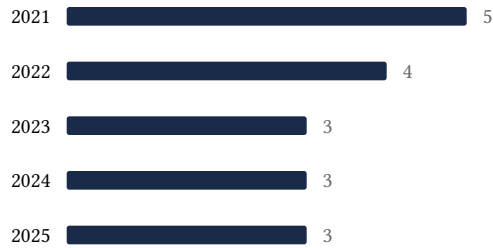
Country	Citing papers
United States	28
United Kingdom	14
Germany	9
Italy	7
China	6
Netherlands	5
Canada	5
Australia	5
Japan	3
Austria	2
France	2
Poland	2

Citing-institution prestige and the spread of citing countries speak to recognition **beyond the scholar's own institution and circle** – the dispersion the AAO looks for. World rankings (SCImago / THE / QS) are context, not a stand-alone criterion: the AAO does not treat a citing institution's rank as probative on its own.

E. Citation Growth Over Time

Distinct citing papers by publication year. Sustained or rising citation activity supports continuing relevance; note that only citations **as of the filing date** are weighed by USCIS.





F. AAO Precedent Considerations

Pre-filing self-check (AAO denial patterns)

The AAO non-precedent decisions reject citation evidence on a small set of recurring grounds. Confirm the petition addresses each before filing:

- Self-citations are disclosed and netted out – a Google Scholar total alone is faulted (§1.1).
- Evidence is per individual article, not a body-of-work aggregate total (§1.2).
- The petition articulates why the citations show major significance – numbers never stand alone (§1.5).
- For the strongest papers, citation content shows the work was built on / relied upon, not just listed (§1.6, §2.2).
- Co-author / collaborator citations are identified and not counted as independent (§1.7).
- Recognition is shown beyond the scholar's own institution and circle (§1.8).
- Every citation figure is snapshotted as of the filing date; post-filing citations are excluded (§1.9).
- Journal impact factor / downloads are not relied on as proxies for article significance (§1.10, §1.12).
- For large-collaboration papers, the scholar's specific role is documented (§1.13).
- Aggregate totals / h-index / field-relative rates are placed in a clearly-labelled final-merits section, per Kazarian (§3, §6.1.7).

Disclaimer

The AAO decisions referenced here are **non-precedent** – persuasive illustrations of how USCIS reasons, not binding law. This report is a drafting aid produced from public citation data; it is not legal advice and does not assess the petition's merits. All analysis must be reviewed by qualified immigration counsel.

G. Citation Evidence Index

Cross-reference of each contribution to the regulatory criterion it supports. Counsel should map these to the petition's exhibit numbers.

Contribution	Core paper	Indep. cites	Supports
Contribution 1	Genome-wide association study identifies novel breast cancer susceptibility loci	14	8 CFR 204.5(h)(3)(v) – Criterion 5
Contribution 2	Breast Cancer Risk Genes-Association Analysis in More than 113,000 Women.	9	8 CFR 204.5(h)(3)(v) – Criterion 5

Contribution	Core paper	Indep. cites	Supports
Contribution 3	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results	7	8 CFR 204.5(h)(3)(v) – Criterion 5