La sintesi degli articoli e le citazioni

A cura di

Domenica Gazineo e Lea Godino
Referenti Formazione e Ricerca SIAN
Riccione
08/05/2024

Why you should read this article:

- To appreciate the use of Microsoft Excel for systematic reviews
- To increase your awareness of all the steps of a systematic review, including importing references and documents, plus documenting the whole process, including intermediate results
- To learn how to create transparent and complete reports for systematic reviews

How to structure Microsoft Excel documents for systematic reviews

Lea Godino

Figure I. The six steps of the Excel method

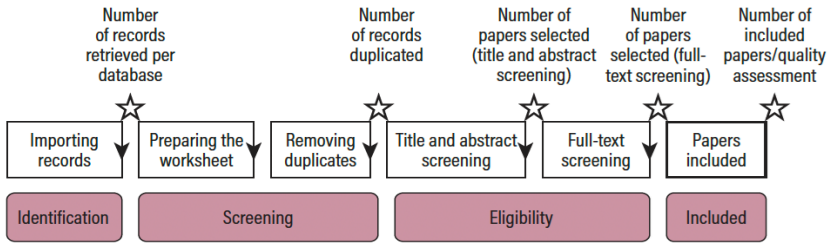

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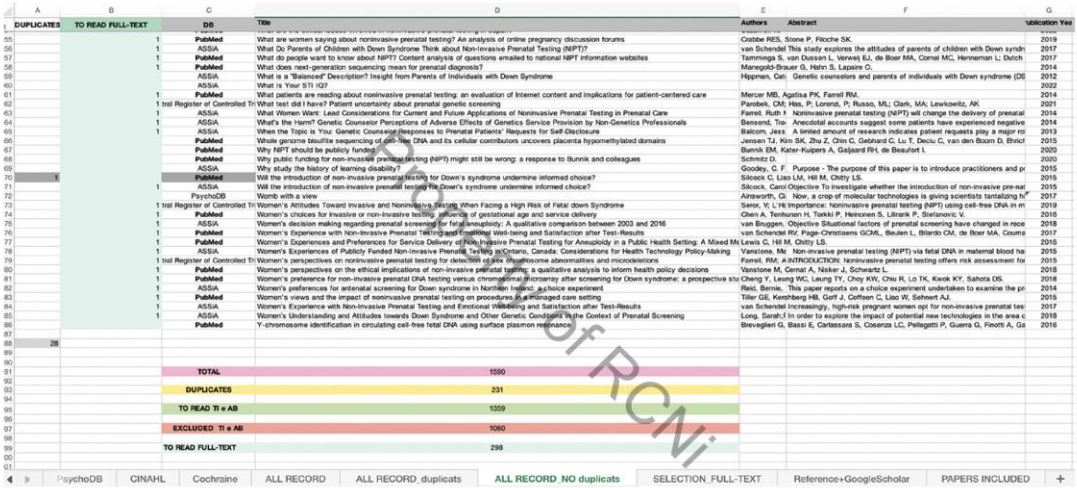
Figure 2. The worksheet after screening the references' titles and abstracts



	A	B	C	D	E	F	G
	DUPLICATES	TO READ FULL-TEXT	DB	Title	Authors	Abstract	Publication Yea
55			1 PubMed	What are women saying about noninvasive prenatal testing? An analysis of online pregnancy discussion forums	Crebbe RES, Stone P, Filoche SK.		2019
56			1 ASSIA	What Do Parents of Children with Down Syndrome Think about Non-Invasive Prenatal Testing (NIPT)?	van Schendel This study explores the attitudes of parents of children with Down syndr		2017
57			1 PubMed	What do people want to know about NIPT? Content analysis of questions emailed to national NIPT information websites	Tammings S, van Dussen L, Verweij EJ, de Boer MA,, Carmel MC, Henneman L; Dutch		2017
58			1 PubMed	What does next-generation sequencing mean for prenatal diagnosis?	Manegold-Brauer G, Hahn S, Lapaire O.		2014
59			ASSIA,	What is a "Balanced" Description? Insight from Parents of Individuals with Down Syndrome	Hippman, Cat Genetic counselors and parents of individuals with Down syndrome (DE		2012
60			ASSIA	What is Your STI IQ?			2022
61			1 PubMed	What patients are reading about noninvasive prenatal testing: an evaluation of Internet content and implications for patient-centered care	Mercer MB, Agatisa PK, Farrell RM,		2014
62			1 trial Register of Controlled Tr	What test did I have? Patient uncertainty about prenatal genetic screening	Parobek, CM; Has, P; Lorenzi, P; Russo, ML; Clark, MA; Lewkowitz, AK		2021
63			1 ASSIA	What Women Want: Lead Considerations for Current and Future Applications of Noninvasive Prenatal Testing in Prenatal Care	Farrell, Ruth M Noninvasive prenatal testing (NIPT) will change the delivery of prenatal		2014
64			1 ASSIA,	What's the Harm? Genetic Counselor Perceptions of Adverse Effects of Genetics Service Provision by Non-Genetics Professionals	Bensend, Tra Anecdotal accounts suggest some patients have experienced negative		2014
65			1 ASSIA,	When the Topic is You: Genetic Counselor Responses to Prenatal Patients' Requests for Self-Disclosure	Balcom, Jess A limited amount of research indicates patient requests play a major rol		2013
66			PubMed	Whole genome bisulfite sequencing of cell-free DNA and its cellular contributors uncovers placenta hypomethylated domains	Jensen TJ, Kim SK, Zhu Z, Chiu C, Gebhard C, Lu T, Deciu C, van den Boom D, Ehrich		2015
67			PubMed	Why NIPT should be publicly funded	Bunnik EM, Kater-Kuipers A, Galjaard RH, de Beaufort I.		2020
68			PubMed	Why public funding for non-invasive prenatal testing (NIPT) might still be wrong: a response to Bunnik and colleagues	Schmitz O.		2020
69			ASSIA,	Why study the history of learning disability?	Goodey, C. F Purpose - The purpose of this paper is to introduce practitioners and pr		2015
70	1		PubMed	Will the introduction of non-invasive prenatal testing for Down's syndrome undermine informed choice?	Silcock C, Liao LM, Hill M, Chitty LS.		2015
71			ASSIA	Will the introduction of non-invasive prenatal testing for Down's syndrome undermine informed choice?	Silcock, Carol Objective To investigate whether the introduction of non-invasive pre-nat		2015
72			1 PsychoDB	Womb with a view	Ainsworth, Cli Now, a crop of molecular technologies is giving scientists tantalizing h		2017
73			1 trial Register of Controlled Tr	Women's Attitudes Toward Invasive and Noninvasive Testing When Facing a High Risk of Fetal down Syndrome	Saror, Y; L Hi Importance: Noninvasive prenatal testing (NIPT) using cell-free DNA in m		2019
74			1 PubMed	Women's choices for invasive or non-invasive testing, efficiency of gestational age and service delivery	Chen A, Tenhunen H, Tonkki P, Heinonen S, Lilrank P, Stefanovic V.		2016
75			1 ASSIA	Women's decision making regarding prenatal screening for fetal aneuploidy: A qualitative comparison between 2003 and 2016	van Bruggen, Objective Situational factors of prenatal screening have changed in rece		2018
76			1 PubMed	Women's Experience with Non-Invasive Prenatal Testing and Emotional Well-being and Satisfaction after Test-Results	van Schendel RV, Page-Christiaens GCML, Beulen L, Bilardo CM, de Boer MA, Coumo		2017
77			1 PubMed	Women's Experiences and Preferences for Service Delivery of Non-Invasive Prenatal Testing for Aneuploidy in a Public Health Setting: A Mixed Me	Lewis C, Hill M, Chitty LS.		2016
78			1 ASSIA,	Women's Experiences of Publicly Funded Non-Invasive Prenatal Testing in Ontario, Canada: Considerations for Health Technology Policy-Making	Vanstone, Me Non-invasive prenatal testing (NIPT) via fetal DNA in maternal blood ha		2015
79			1 trial Register of Controlled Tr	Women's perspectives on noninvasive prenatal testing for detection of sex chromosome abnormalities and microdeletions	Farrell, RM; A INTRODUCTION: Noninvasive prenatal testing offers risk assessment for		2015
80			1 PubMed	Women's perspectives on the ethical implications of non-invasive prenatal testing: a qualitative analysis to inform health policy decisions	Vanstone M, Cernat A, Nisker J, Schwartz L.		2018
81			1 PubMed	Women's preference for non-invasive prenatal DNA testing versus chromosomal microarray after screening for Down syndrome: a prospective stu	Cheng Y, Leung WC, Leung TY, Choy KW, Chiu R, Lo TK, Kwok KY, Sahota DS.		2018
82			1 ASSIA	Women's preferences for antenatal screening for Down syndrome in Northern Ireland: a choice experiment	Reid, Bernie, This paper reports on a choice experiment undertaken to examine the pr		2014
83			1 PubMed	Women's views and the impact of noninvasive prenatal testing on processes in a managed care setting	Tiller GE, Kerenberg HB, Goff J, Coffeen C, Liao W, Seheert AJ.		2015
84			1 ASSIA,	Women's Experience with Non-Invasive Prenatal Testing and Emotional Well-being and Satisfaction after Test-Results	van Schendel Increasingly, high-risk pregnant women opt for non-invasive prenatal tes		2017
85			1 ASSIA	Women's Understanding and Attitudes towards Down Syndrome and Other Genetic Conditions in the Context of Prenatal Screening	Long, Sarah; I In order to explore the impact of potential new technologies in the area c		2018
86			PubMed	Y-chromosome identification in circulating cell-free fetal DNA using surface plasmon resonance	Brevegliari G, Bassi E, Carbassas S, Cosenza LC, Pellegatti P, Guerra G, Finotti A, Ga		2016
87	28						
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89				TOTAL 1590			
90				DUPLICATES 231			
91				TO READ TI e AB 1359			
92				EXCLUDED TI e AB 1060			
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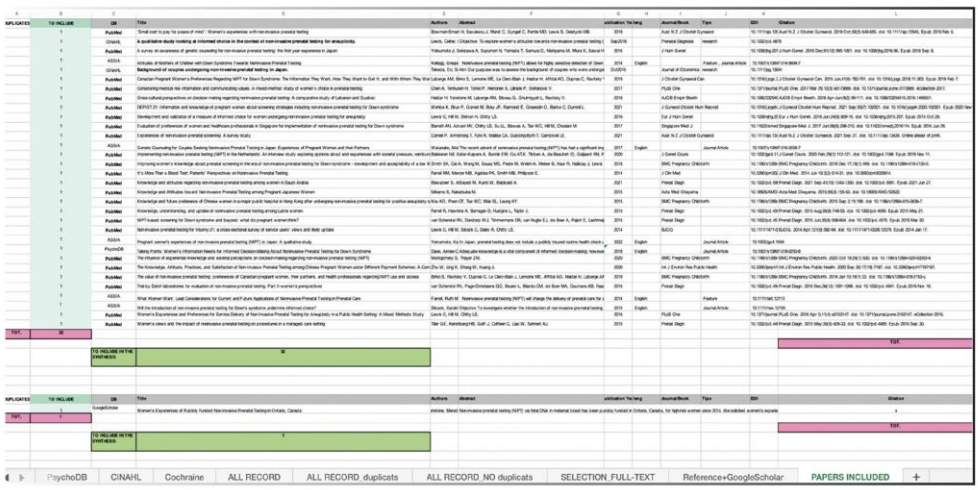
Lea Godino

Figure 3. The final list of articles to include in the systematic review



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Gazineo D, Godino L, Savini S, Stefanizzi G, Scarpo E, Del Pin M, Canzi M, Zito MP, Fabbri C. [The COVID-19 emergency management in Nephrology: a cross-sectional survey on the procedures management to deal with the pandemic]. G Ital Nefrol. 4(8) (2022) pp. 85–93.

Giornale Italiano di Nefrologia

La gestione dell'emergenza COVID-19 in ambito nefrologico: i risultati di un'indagine trasversale sulla gestione delle procedure atte a fronteggiare la pandemia

Articoli originali

Domenica Gazineo[1], Lea Godino[1], Serenella Savini[2], Stefanizzi Grazia[1], Emiliana Scarpo[2], Marco Del Pin[3], Canzi Mara[4], Maria Pia Zito[4], Cinzia Fabbri[1]

1 IRCCS Azienda Ospedaliera Universitaria di Bologna (BO), Italia
2 ASL Roma 4 Civitavecchia (Roma), Italia
3 ASUSF Ospedale di Palmanova (UDINE), Italia
4 Società Italiana Infermieri Area Nefrologica (SIAN), Italia

Domenica Gazineo

Corrispondenza a:
Dott.ssa Domenica Gazineo
IRCCS Azienda Ospedaliero Universitaria di Bologna via Albertoni, 15
40138 Bologna
Tel. 3385436783
Mail: domenica.gazineo3@unibo.it

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ALCUNI ESEMPI

European Journal of Human Genetics (2016) 24, 496–503

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REVIEW

Impact of presymptomatic genetic testing on young adults: a systematic review

Lea Godino*[,1,2], Daniela Turchetti[1], Leigh Jackson[2], Catherine Hennessy[2] and Heather Skirton[2]

Presymptomatic and predictive genetic testing should involve a considered choice, which is particularly true when testing is undertaken in early adulthood. Young adults are at a key life stage as they may be developing a career, forming partnerships and potentially becoming parents: presymptomatic testing may affect many facets of their future lives. The aim of this integrative systematic review was to assess factors that influence young adults' or adolescents' choices to have a presymptomatic genetic test and the emotional impact of those choices. Peer-reviewed papers published between January 1993 and December 2014 were searched using eight databases. Of 3373 studies identified, 29 were reviewed in full text: 11 met the inclusion criteria. Thematic analysis was used to identify five major themes: period before testing, experience of genetic counselling, parental involvement in decision-making, impact of test result communication, and living with genetic risk. Many participants grew up with little or no information concerning their genetic risk. The experience of genetic counselling was either reported as an opportunity for discussing problems or associated with feelings of disempowerment. Emotional outcomes of disclosure did not directly correlate with test results: some mutation carriers were relieved to know their status, however, the knowledge they may have passed on the mutation to their children was a common concern. Parents appeared to have exerted pressure on their children during the decision-making process about testing and risk reduction surgery. Health professionals should take into account all these issues to effectively assist young adults in making decisions about presymptomatic genetic testing.
European Journal of Human Genetics (2016) **24,** 496–503; doi:10.1038/ejhg.2015.153; published online 15 July 2015

INTRODUCTION

Presymptomatic and predictive genetic testing are available for a number of heritable genetic disorders including hereditary cancer syndromes, inherited cardiac conditions and neurodegenerative genetic disorders.[1] The terms 'presymptomatic' and 'predictive' genetic testing refer to the possibility of detecting a genetic mutation that causes a particular condition before the presentation of symptoms. The first term generally refers to those diseases in which a positive test result will inevitably lead to the development of the disease later in life (eg, Huntington disease (HD)); the second term refers to a broader range of diseases in which the risk for a disorder is increased but without necessarily implying any degree of certainty (eg, hereditary breast and ovarian cancer (HBOC)). However, these terms are often used in a broadly interchangeable manner. A substantial difference is that cancer disorders can be monitored through a surveillance protocol or prevented by surgical intervention, while no prevention is currently available for diseases such as HD or cerebellar ataxia. Therefore, the choice to undergo a presymptomatic test for disorders with incomplete penetrance and where there are preventive measures could have a highly different psychological and social impact when compared with testing for disorders with complete penetrance and no preventive options, particularly in young adults. In this review, the term 'presymptomatic' will be used to indicate both predictive and presymptomatic tests, but the different impact will be considered whenever appropriate.

A presymptomatic diagnosis of a serious genetic illness can have a profound impact on the person and family and should be managed using an individualized counselling process.[1] Presymptomatic genetic testing of minors (under the age of 18 years) is not usually recommended unless effective clinical actions are available.[2–4] Generally, there are three key arguments against presymptomatic genetic testing in adolescents or young people: that it (1) fails to respect the future autonomy of the young person; (2) breaches confidentiality; and (3) may cause psychosocial harms.[5]

The age at which young people should undergo presymptomatic genetic testing for adult-onset disorder is a matter of debate.[2–5] Key challenges have to be faced during the transition from adolescence to adulthood, such as marriage, finishing education, beginning full-time employment and becoming a parent, and the impact of testing may affect, and be affected by each of these events. In the light of the above-mentioned issues, it would be appropriate to ask what health information and counselling young adults need to make prudent decisions about genetic testing. The purpose of this systematic review was therefore to systematically identify and analyse factors influencing young adults' or adolescents' choices to have a presymptomatic test and the emotional impact of those choices.

MATERIALS AND METHODS

Design

A systematic review is a method of amassing, assessing and synthesizing a body of evidence on a particular topic.[6] This systematic review was conducted in accordance with the Centre for Reviews and Dissemination methods for undertaking reviews in health care[6] with the aim of assessing which factors influence young adults' or adolescents' choices to have a presymptomatic test for a genetic condition and the emotional impact of those choices.

1 Evans JP, Skrzynia C, Burke W: The complexities of predictive genetic testing. *BMJ* 2001; **322**: 1052–1056.
2 MacLeod R, Beach A, Henriques S, Knopp J, Nelson K, Kerzin-Storrar L: Experiences of predictive testing in young people at risk of Huntington's disease, familial cardiomyopathy or hereditary breast and ovarian cancer. *Eur J Hum Genet* 2013; **22**: 396–401.
3 Bonfy P, Goffin T, Ng H, Dierickx K: Predictive genetic testing in minors for adult-onset genetic diseases. *Mt Sinai J Med* 2008; **75**: 287–296.
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10 Braun V, Clarke V: Using thematic analysis in psychology. *Qual Res Psychol* 2008; **3**: 77–101.
11 MacLeod R, Beach A, Henriques S, Knopp J, Nelson K, Kerzin-Storrar L: Experiences of predictive testing in young people at risk of Huntington's disease, familial cardiomyopathy or hereditary breast and ovarian cancer. *Eur J Hum Genet EJHG* 2014; **22**: 396–401.
12 Duncan RE, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatycki MB: "Holding your breath": Interviews with young people who have undergone predictive genetic testing for Huntington disease. *Am J Med Genet Part A* 2007; **143A**: 1984–1989.

important to consider the influence of the specific disease considered: the perception and experience of harms and benefits from the test result for a potentially treatable condition (such as BRCA and FAP, etc) may not be the same as for conditions for which there are no preventive treatment or cure (such as HD).

A potential limitation of this systematic review is that all the papers analysed are based on studies conducted in only four countries with similar British historical and cultural legacies, thus the findings may not generalize to other countries with different sociocultural backgrounds, supporting the need for further studies in other contexts. On the other hand, the papers analysed spanned across several diseases, while considering similar age ranges, thus providing a comprehensive overview of how young adults deal with genetic testing overall and according to the specific disease.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

ACKNOWLEDGEMENTS

LG is supported by the Grant from Regione Emilia-Romagna 'Diagnostics advances in hereditary breast cancer (DIANE)' (PRUa1GR-2012-001).

13 Duncan RE, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatycki MB: The challenge of developmentally appropriate care: predictive genetic testing in young people for familial adenomatous polyposis. *Fam Cancer* 2010; **9**: 27–35.
14 Marmar L, de Souza AN, Loiselle CG, Wong N: Experience of BRCA1/2 mutation-negative young women from families with hereditary breast and ovarian cancer: a qualitative study. *Hered Cancer Clin Pract* 2013; **11**: 14.
15 Mand C, Gillam L, Duncan RE, Delatycki MB: "It was the missing piece": adolescent experiences of predictive genetic testing for adult-onset conditions. *Genet Med* 2013; **15**: 643–649.
16 Patenaude AF, Tung N, Ryan PD, Ellisen LW, Hewitt L, Schneider KA *et al*: Young adult daughters of BRCA1/2 positive mothers. What do they know about hereditary cancer and how much do they worry? *Psychooncology* 2013; **22**: 2024–2031.
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ALCUNI ESEMPI

Nursing & Health Sciences

Nursing and Health Sciences (2013), **15**, 15–21

Research Article

Genetic counseling: A survey to explore knowledge and attitudes of Italian nurses and midwives

Lea Godino, RN,[1,2] Daniela Turchetti, MD[1] and Heather Skirton, PhD[2]
[1]*Medical Genetic Unit, University of Bologna, Bologna, Italy and* [2]*Faculty of Health, Education and Society, Plymouth University, Plymouth, UK*

Abstract In the past, genetic services were delivered to a limited number of families with rare conditions. However, genomics is now being applied to both inherited and common diseases in a range of healthcare settings, and there is a greater need for nurses to understand the basic concepts of genetic health care. The aim of this cross-sectional survey was to explore the understanding and attitudes of Italian nurses toward genetic health care. A questionnaire was completed by 102 nurses and midwives (85% response rate). Of these, 61% believed that genetic counseling was only an informative and advisory process, and 53.9% could not specify to whom the counseling was aimed. When asked to identify nurses' role in genetic health care, 62% of the respondents believed they had no role, although 28% believed that nurses could provide information, support, and counseling. These findings indicate that nurses have only partial knowledge of the issues surrounding genetic health care. To prepare nurses for the post-genomic era, improved genetic education at the undergraduate and postgraduate levels is required.

Key words genetic counselling, genetics, Italy, midwives, nurses, survey.

INTRODUCTION

In the past, medical genetics was a small specialty, delivering services to a limited number of families with conditions caused by a chromosomal abnormality or genetic mutation. Individually, each of these conditions is relatively rare; however, when viewed as a group, it is clear that genetic conditions do affect a significant number of people in the general population. Nurses have always been involved in offering care to individuals who are affected by one of a large number of genetic conditions, but this situation is changing. In the past, access to specialist genetic services and genetic testing was generally not available to patients with concerns about the genetic component of more common diseases, such as coronary heart disease and diabetes. Developments in technology have enabled the detection of genetic contributions to common diseases (Collins, 2004) and the identification of inherited subsets of diseases, such as diabetes (Shepherd *et al.*, 2001). As a result, the study of genetics has become progressively more important to healthcare providers in Europe, where an estimated 30 million people now suffer from diseases with a genetic component (Cassiman, 2005). Nowadays, the number of people requiring genetic counseling has increased due to technological progress and molecular research. In addition, there have been some sociocultural changes, such as increasing maternal age for first pregnancies and medically-assisted procreation (Bricarelli *et al.*, 2001). In view of these changes, genetic counseling activities need to be offered by a wider group of health professionals, so as to provide genetic information, education, and support to patients and their families with current and/or future genetic problems (Frazier *et al.*, 2004).

In an article focused on nursing education, Lea (2002) explains not only basic genetics, but also the ways in which nurses can integrate a genetic focus into practice. This is important to enable nurses to help patients and their families understand genetic characteristics, genetic transmission, and how the expression of disease varies depending on environment. Lea (2002) therefore recommends that an initial nursing assessment should include taking a family history, to enable the nurse to identify a potential pattern of inheritance (e.g. Mendelian pattern or identification of a multifactorial condition), as this might have implications for the ongoing nursing and medical management of the family.

In many countries, genetic nurses are central to genetic health care. Genetic nurses currently work in many countries, including the UK, Japan, and the USA (Skirton *et al.*, 2010a). There are diverse groups of professionals (including experts in clinical genetics, biologists, and psychologists) that see the need to integrate genetic knowledge with nursing science (Williams *et al.*, 2004). Nurses need to use genetic information in care settings, such as cardiovascular nursing, geriatric nursing, oncology nursing, public health, nursing education, and primary care (Jenkins *et al.*, 2001). In fact, in a survey by

Correspondence address: Lea Godino, University of Bologna, UO Genetica Medica, Policlinico Sant'Orsola-Malpighi, via Massarenti, 9, Bologna 40138, Italy. Email: leagodino@gmail.com
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Table 3. Role of nurses and midwives in providing genetic counseling

	Providing information, counseling, and support	Collecting data	Minimal role	No role
Role of nurses in providing genetic counseling	28.0% ($n = 29$)	3.0% ($n = 3$)	7.0% ($n = 7$)	62% ($n = 63$)
Role of midwives in providing genetic counseling	39.0% ($n = 40$)	2.0% ($n = 2$)	15.0% ($n = 15$)	44.0% ($n = 45$)

for mainstream health care, and genetics education was not needed to prepare nurses for practice. In an international survey of nursing leaders in 10 countries from six continents, Kirk *et al.* (2011) also discovered that there were existing challenges to integrating genetics into nursing in all the countries studied; these included identifying nursing competences in genetics to integrating genetics into the nursing curricula. The authors call for more support by nurses at senior levels in government, regulatory bodies, and education to facilitate change, but do not specifically cite nurse managers at the institutional level as key personnel in this regard. As the low priority of genetics in nursing appears to be a global issue, it is unsurprising that nurses in Italy do not recognize the relevance of genetics to their practice.

The present study was a small study of nurses and midwives working in one city, and in departments where referral to genetic services is most likely (perinatal care). Although the response rate was very good (85%), further research is needed to establish whether the views and knowledge of nurses in this study are reflected across Italy and in different clinical contexts. We acknowledge the weaknesses of the study; one being the lack of a pilot study, and another, the use of invalidated tools. However, we were unable to find any previously used tools that would have served the purpose of surveying nurses on this topic.

Conclusion

Genetic counseling is not only a transmission of information, but a process in which patients' emotional and psychological situations are considered and supported (Freshwater, 2003). In view of the findings, there appears to be a need to include more genetics material in nurse training courses, in particular with respect to genetic counseling and medical genetics issues. Nurses who work closely with patients and their families can then play a more effective role in supporting those with genetic conditions.

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CONTRIBUTIONS

Study Design: LG, DT.
Data Collection and Analysis: LG, DT.
Manuscript Writing: LG, HS, DT.

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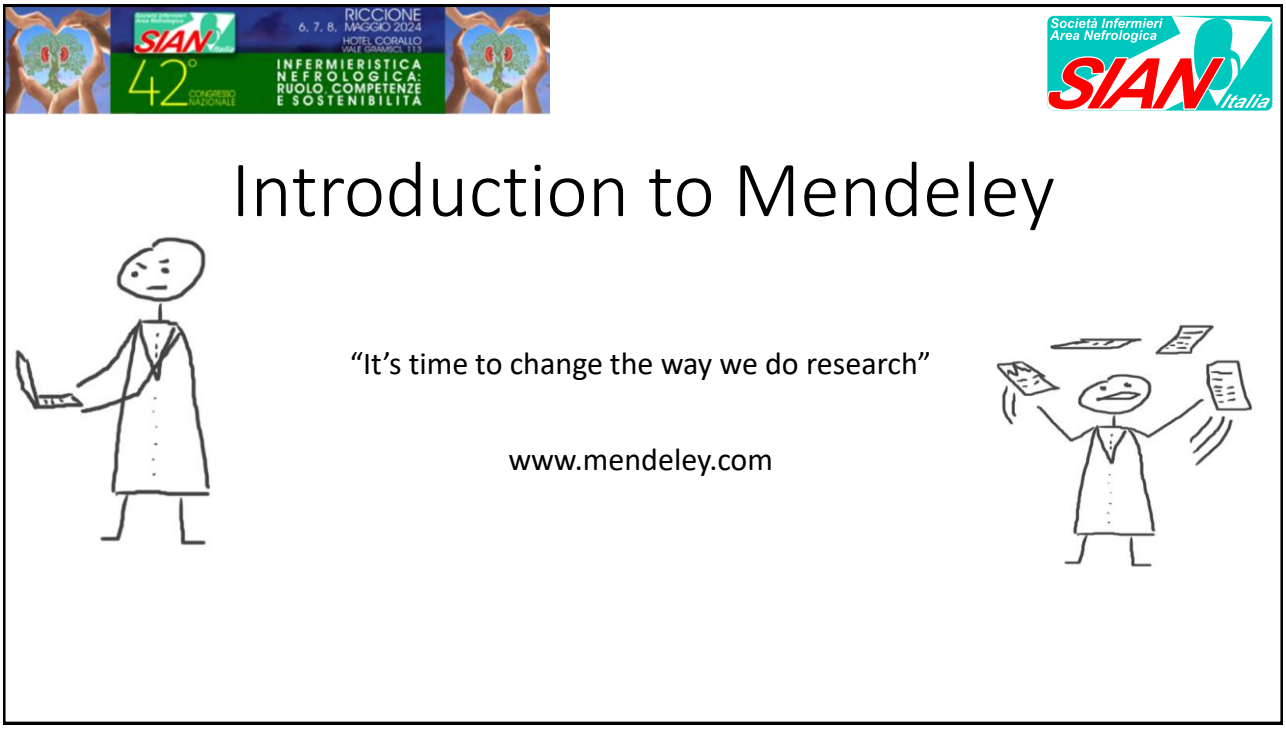

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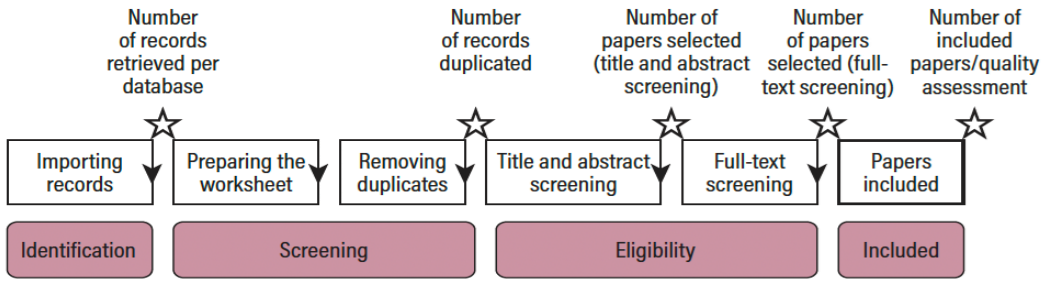

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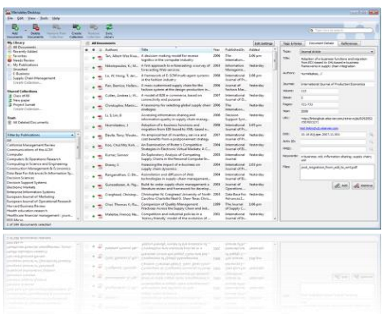

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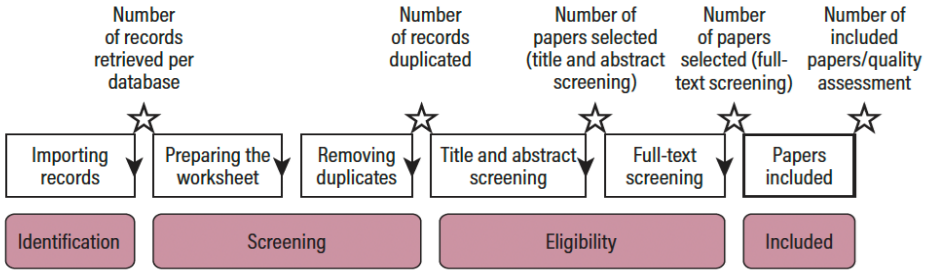

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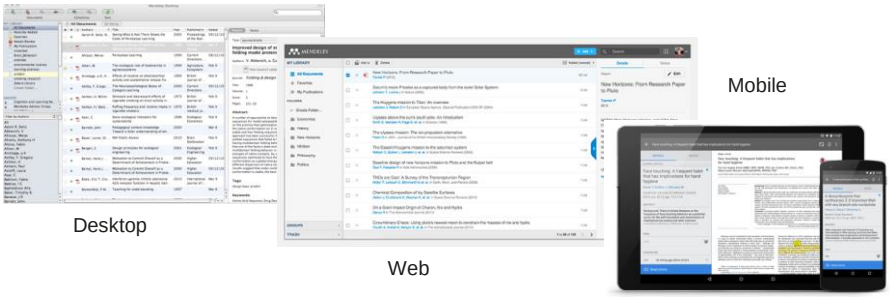

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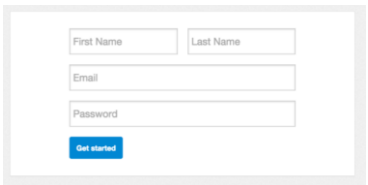

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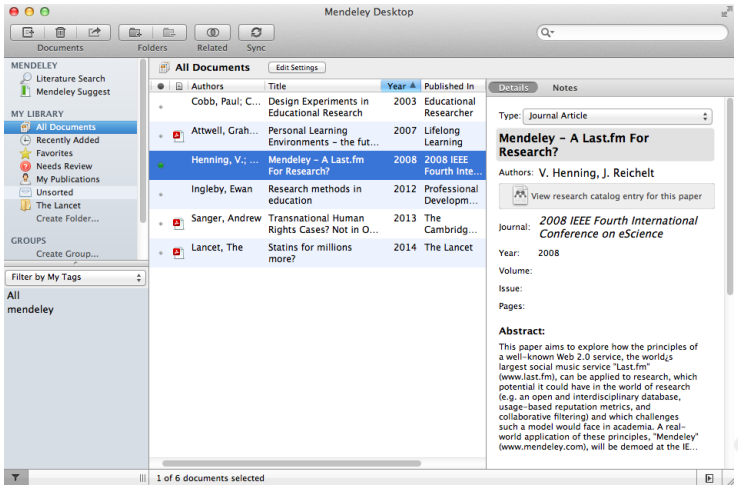

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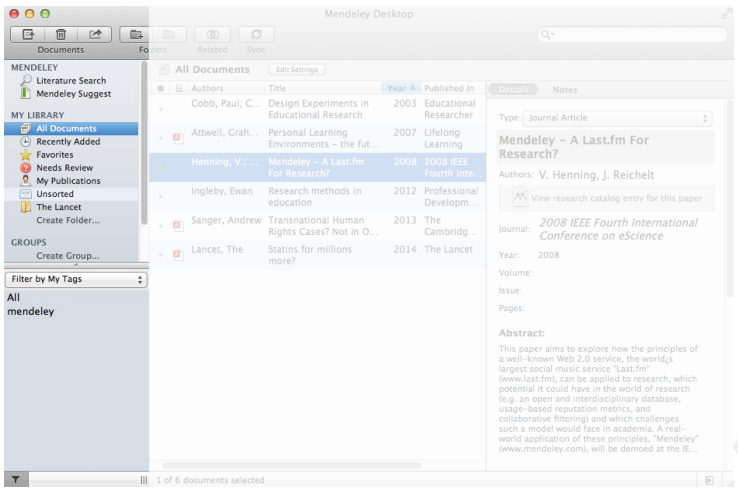

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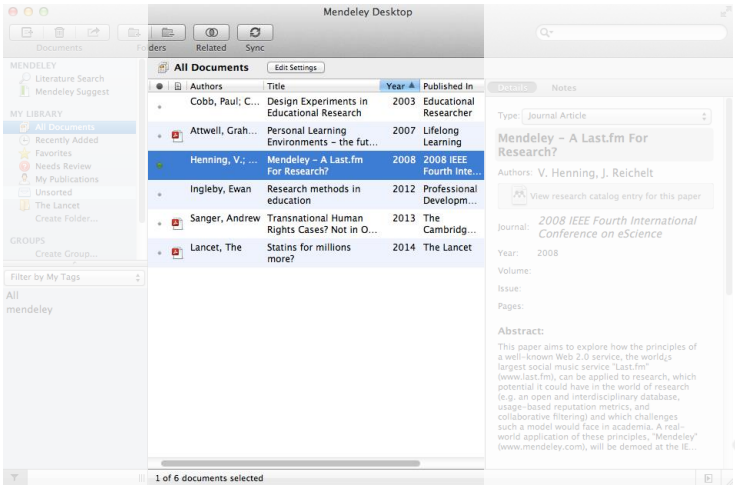

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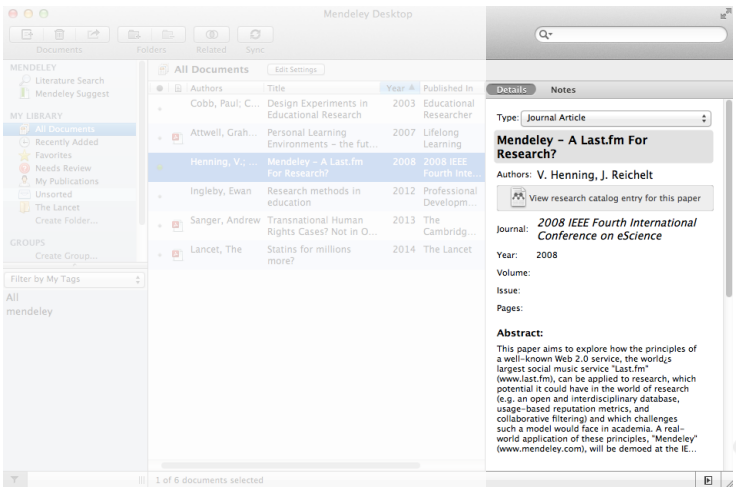

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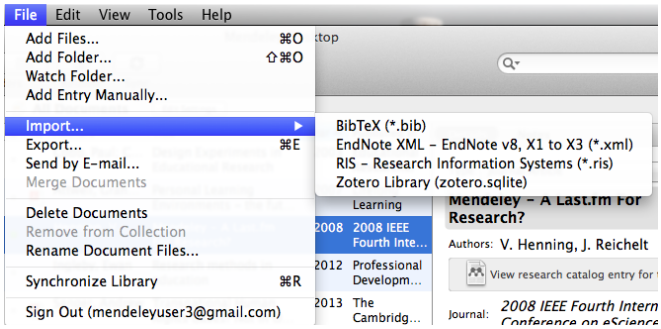

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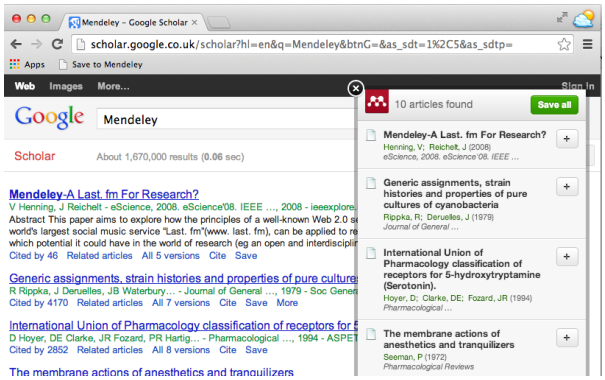

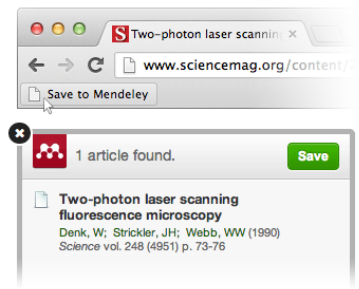

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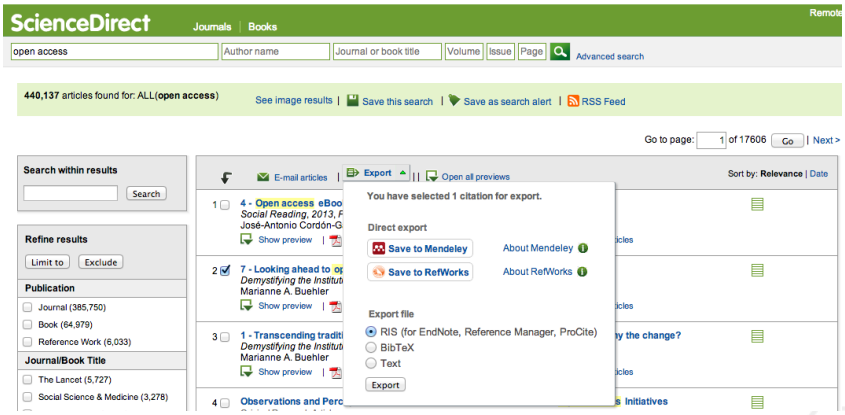

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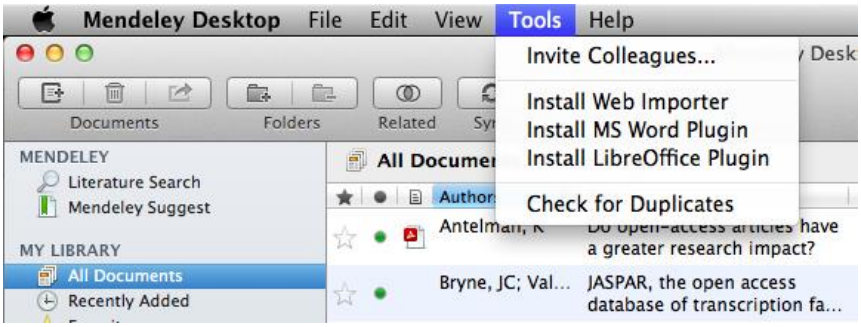

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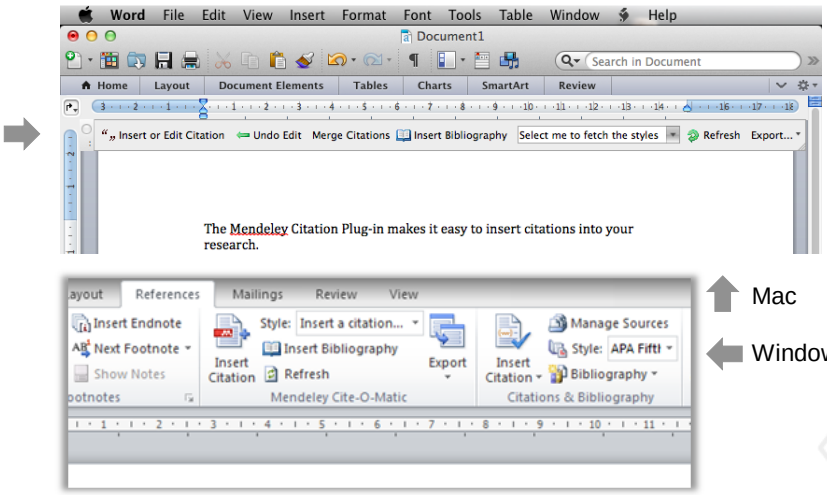

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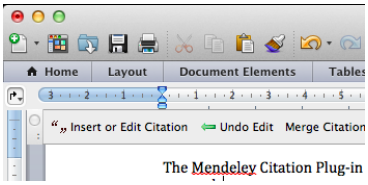

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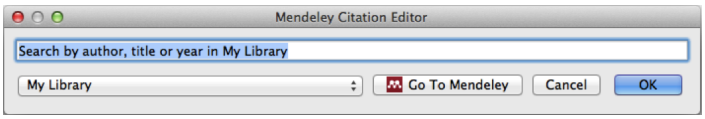

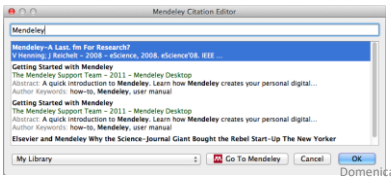

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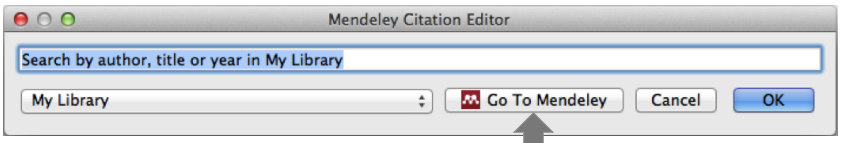

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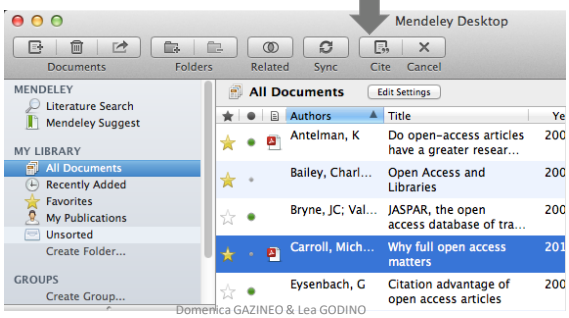

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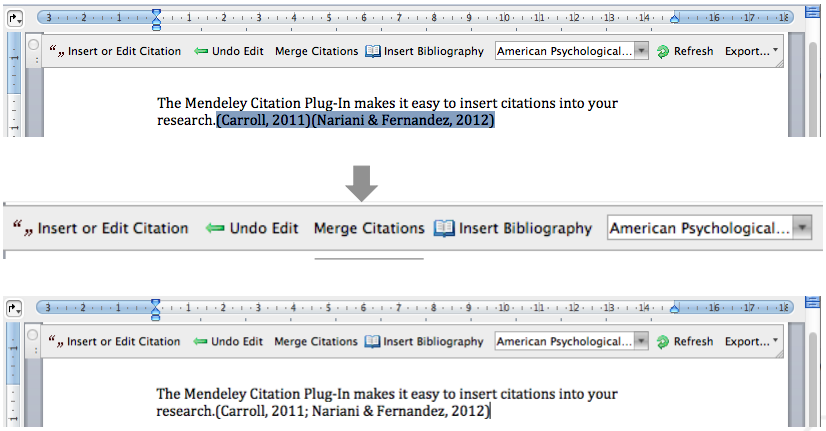

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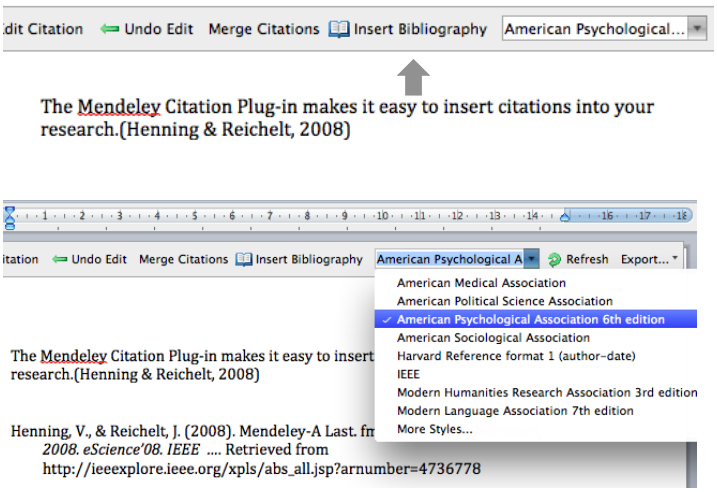

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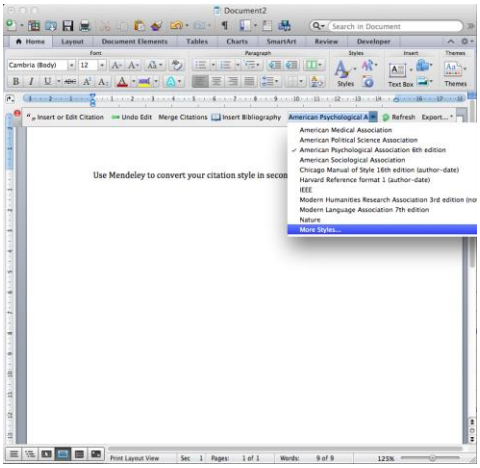

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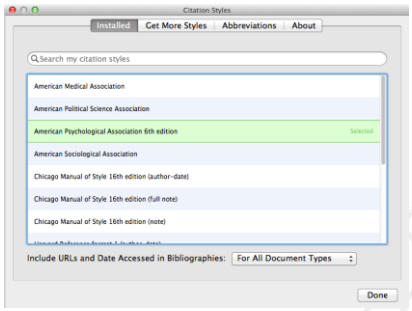

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