Sylvia A Metcalfe

List of Publications by Year
in descending order

87 papers	2,647 citations	159585 30 h-index	223800 46 g-index
87 all docs	87 docs citations	87 times ranked	2506 citing authors

#	Article	IF	Citations
1	The expectations and realities of nutrigenomic testing in australia: A qualitative study. Health Expectations, 2021, 24, 670-686.	2.6	3
2	From Expectations to Experiences: Consumer Autonomy and Choice in Personal Genomic Testing. AJOB Empirical Bioethics, 2020, 11, 63-76.	1.6	9
3	Editorial: Educating Health Professionals in Genomic Medicine: Evidence-Based Strategies and Approaches. Frontiers in Genetics, 2020, 11, 696.	2.3	0
4	Human Genetics Society of Australasia Position Statement: Online DNA Testing. Twin Research and Human Genetics, 2020, 23, 256-258.	0.6	2
5	Preparing Medical Specialists for Genomic Medicine: Continuing Education Should Include Opportunities for Experiential Learning. Frontiers in Genetics, 2020, 11, 151.	2.3	45
6	Development of an Evidence-Based, Theory-Informed National Survey of Physician Preparedness for Genomic Medicine and Preferences for Genomics Continuing Education. Frontiers in Genetics, 2020, 11, 59.	2.3	20
7	Australiansâ€™ views and experience of personal genomic testing: survey findings from the Genioz study. European Journal of Human Genetics, 2019, 27, 711-720.	2.8	14
8	Personal genomic testing for nutrition and wellness in Australia: A content analysis of online information. Nutrition and Dietetics, 2019, 76, 263-270.	1.8	7
9	Readiness of clinical genetic healthcare professionals to provide genomic medicine: An Australian census. Journal of Genetic Counseling, 2019, 28, 367-377.	1.6	29
10	Lessons learnt from implementing change in newborn bloodspot screening processes over more than a decade: Midwives, genetics and education. Midwifery, 2019, 79, 102542.	2.3	3
11	Preparing Medical Specialists to Practice Genomic Medicine: Education an Essential Part of a Broader Strategy. Frontiers in Genetics, 2019, 10, 789.	2.3	50
12	Ensuring Best Practice in Genomic Education and Evaluation: A Program Logic Approach. Frontiers in Genetics, 2019, 10, 1057.	2.3	17
13	Australiansâ€™ perspectives on support around use of personal genomic testing: Findings from the Genioz study. European Journal of Medical Genetics, 2019, 62, 290-299.	1.3	17
14	Genetic counselling, patient education, and informed decision-making in the genomic era. Seminars in Fetal and Neonatal Medicine, 2018, 23, 142-149.	2.3	34
15	FMR1 allele size distribution in 35,000 males and females: a comparison of developmental delay and general population cohorts. Genetics in Medicine, 2018, 20, 1627-1634.	2.4	23
16	Fragile X population carrier screening. Genetics in Medicine, 2018, 20, 1091-1092.	2.4	5
17	Australiansâ€™ views on personal genomic testing: focus group findings from the Genioz study. European Journal of Human Genetics, 2018, 26, 1101-1112.	2.8	14
18	Informed decision making and psychosocial outcomes in pregnant and nonpregnant women offered population fragile X carrier screening. Genetics in Medicine, 2017, 19, 1346-1355.	2.4	20

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19	Clinical audit of genetic testing and referral patterns for fragile X and associated conditions. American Journal of Medical Genetics, Part A, 2016, 170, 1439-1449.	1.2	7
20	â€œIt gives them more optionsâ€: preferences for preconception genetic carrier screening for fragile X syndrome in primary healthcare. Journal of Community Genetics, 2016, 7, 159-171.	1.2	17
21	Experiences of prenatal diagnosis and decisionâ€making about termination of pregnancy: A qualitative study. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2016, 56, 605-613.	1.0	46
22	Responsible implementation of expanded carrier screening. European Journal of Human Genetics, 2016, 24, e1-e12.	2.8	240
23	Outcomes of a randomised controlled trial of a complex genetic counselling intervention to improve family communication. European Journal of Human Genetics, 2016, 24, 356-360.	2.8	55
24	Detection of skewed X-chromosome inactivation in Fragile X syndrome and X chromosome aneuploidy using quantitative melt analysis. Expert Reviews in Molecular Medicine, 2015, 17, e13.	3.9	12
25	A mixed methods study of age at diagnosis and diagnostic odyssey for Duchenne muscular dystrophy. European Journal of Human Genetics, 2015, 23, 1294-1300.	2.8	39
26	Evidence linking FMR1 mRNA and attentional demands of stepping and postural control in women with the premutation. Neurobiology of Aging, 2015, 36, 1400-1408.	3.1	10
27	Novel methylation markers of the dysexecutive-psychiatric phenotype in FMR1 premutation women. Neurology, 2015, 84, 1631-1638.	1.1	32
28	Development of a fragile <scp>X</scp> syndrome (<scp>FXS</scp>) knowledge scale: towards a modified multidimensional measure of informed choice for <scp>FXS</scp> population carrier screening. Health Expectations, 2015, 18, 69-80.	2.6	12
29	Carrier testing in children and adolescents. European Journal of Medical Genetics, 2015, 58, 659-667.	1.3	11
30	Analyzing communication in genetic consultationsâ€”A systematic review. Patient Education and Counseling, 2015, 98, 15-33.	2.2	42
31	A mixed methods exploration of familiesâ€™ experiences of the diagnosis of childhood spinal muscular atrophy. European Journal of Human Genetics, 2015, 23, 575-580.	2.8	32
32	Preferences for results from genomic microarrays: comparing parents and health care providers. Clinical Genetics, 2015, 87, 21-29.	2.0	18
33	Population-based carrier screening for cystic fibrosis: a systematic review of 23 years of research. Genetics in Medicine, 2014, 16, 207-216.	2.4	72
34	Impaired response inhibition is associated with selfâ€reported symptoms of depression, anxiety, and ADHD in female FMR1 premutation carriers. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 41-51.	1.7	48
35	Improving family communication after a new genetic diagnosis: a randomised controlled trial of a genetic counselling intervention. BMC Medical Genetics, 2014, 15, 33.	2.1	26
36	A qualitative exploration of mothersâ€™ and fathersâ€™ experiences of having a child with Klinefelter syndrome and the process of reaching this diagnosis. European Journal of Human Genetics, 2014, 22, 18-24.	2.8	32

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37	Prenatal Î²â€thalassemia carrier screening in Australia: healthcare professionals' perspectives of clinical practice. Prenatal Diagnosis, 2014, 34, 246-250.	2.3	8
38	Availability of treatment drives decisions of genetic health professionals about disclosure of incidental findings. European Journal of Human Genetics, 2014, 22, 1225-1228.	2.8	13
39	Age and CGG-repeat length are associated with neuromotor impairments in at-risk females with the FMR1 premutation. Neurobiology of Aging, 2014, 35, 2179.e7-2179.e13.	3.1	21
40	Exploring inhibitory deficits in female premutation carriers of fragile X syndrome: Through eye movements. Brain and Cognition, 2014, 85, 201-208.	1.8	27
41	â€œHe didnâ€™t say that thalassaemia might come upâ€•â€” Î²-thalassaemia carriersâ€™ experiences and attitudes. Journal of Community Genetics, 2013, 4, 223-232.	1.2	6
42	â€œIt's about having the choiceâ€• Stakeholder perceptions of populationâ€based genetic carrier screening for fragile X syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 48-58.	1.2	25
43	Cognitive-motor interference during postural control indicates at-risk cerebellar profiles in females with the FMR1 premutation. Behavioural Brain Research, 2013, 253, 329-336.	2.2	27
44	Considerations for Reporting Genome Results to Patients. Journal of Paediatrics and Child Health, 2013, 49, 82-82.	0.8	0
45	Why Do People Choose Not to Have Screening for Hemochromatosis?. Genetic Testing and Molecular Biomarkers, 2013, 17, 21-24.	0.7	1
46	Fragile Xâ€“related element 2 methylation analysis may provide a suitable option for inclusion of fragile X syndrome and/or sex chromosome aneuploidy into newborn screening: a technical validation study. Genetics in Medicine, 2013, 15, 290-298.	2.4	19
47	Cascade carrier testing after a child is diagnosed with cystic fibrosis through newborn screening: investigating why most relatives do not have testing. Genetics in Medicine, 2013, 15, 533-540.	2.4	22
48	Current Practice and Attitudes of Australian Obstetricians Toward Population-Based Carrier Screening for Inherited Conditions. Twin Research and Human Genetics, 2013, 16, 601-607.	0.6	22
49	Key Informantsâ€™ Perspectives of Implementing Chromosomal Microarrays Into Clinical Practice in Australia. Twin Research and Human Genetics, 2013, 16, 833-839.	0.6	10
50	â€œTaking Its Tollâ€• The Challenges of Working in Fetal Medicine. Birth, 2013, 40, 52-60.	2.2	22
51	An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice. European Journal of Human Genetics, 2012, 20, 825-830.	2.8	48
52	ironXS: high-school screening for hereditary haemochromatosis is acceptable and feasible. European Journal of Human Genetics, 2012, 20, 505-509.	2.8	7
53	Fragile X population carrier screening. Genetics in Medicine, 2012, 14, 350-350.	2.4	3
54	Carrier screening in preconception consultation in primary care. Journal of Community Genetics, 2012, 3, 193-203.	1.2	40

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55	A case for cystic fibrosis carrier testing in the general population. Medical Journal of Australia, 2011, 194, 208-209.	1.7	17
56	Development of a questionnaire for evaluating genetics education in general practice. Journal of Community Genetics, 2010, 1, 175-183.	1.2	8
57	â€œTesting Times, Challenging Choicesâ€• An Australian Study of Prenatal Genetic Counseling. Journal of Genetic Counseling, 2010, 19, 22-37.	1.6	32
58	â€œIt's Challenging on a Personal Levelâ€â€”Exploring the â€˜Lived Experienceâ€™ of Australian and Canadian Prenatal Genetic Counselors. Journal of Genetic Counseling, 2010, 19, 640-652.	1.6	12
59	Uptake of carrier testing in families after cystic fibrosis diagnosis through newborn screening. European Journal of Human Genetics, 2010, 18, 1084-1089.	2.8	36
60	Carrier screening for Beta-thalassaemia: a review of international practice. European Journal of Human Genetics, 2010, 18, 1077-1083.	2.8	174
61	A systematic review of population screening for fragile X syndrome. Genetics in Medicine, 2010, 12, 396-410.	2.4	85
62	â€œIt's something I need to considerâ€• Decisions about carrier screening for fragile X syndrome in a population of nonâ€pregnant women. American Journal of Medical Genetics, Part A, 2009, 149A, 2731-2738.	1.2	32
63	The Importance of Program Evaluation: How Can it be Applied to Diverse Genetics Education Settings?. Journal of Genetic Counseling, 2008, 17, 170-179.	1.6	21
64	Impact of a Genetic Diagnosis of a Mitochondrial Disorder 5â€“17 Years After the Death of an Affected Child. Journal of Genetic Counseling, 2008, 17, 261-273.	1.6	17
65	â€˜It is not in my worldâ€™: an exploration of attitudes and influences associated with cystic fibrosis carrier screening. European Journal of Human Genetics, 2008, 16, 435-444.	2.8	51
66	Disclosing Genetic Research Results After Death of Pediatric Patients. JAMA - Journal of the American Medical Association, 2008, 300, 1693.	7.4	11
67	A model for offering carrier screening for fragile X syndrome to nonpregnant women: results from a pilot study. Genetics in Medicine, 2008, 10, 525-535.	2.4	51
68	The missing element: consanguinity as a component of genetic risk assessment. Genetics in Medicine, 2008, 10, 612-620.	2.4	13
69	A model for the development of genetics education programs for health professionals. Genetics in Medicine, 2007, 9, 451-457.	2.4	37
70	Population genetic screening. Australian Family Physician, 2007, 36, 794-800.	0.5	2
71	Genetics and blood--haemoglobinopathies and clotting disorders. Australian Family Physician, 2007, 36, 812-9.	0.5	8
72	It's â€˜back to schoolâ€™ for genetic screening. European Journal of Human Genetics, 2006, 14, 384-389.	2.8	18

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73	Evaluation of a decision aid for prenatal testing of fetal abnormalities: a cluster randomised trial [ISRCTN22532458]. BMC Public Health, 2006, 6, 96.	2.9	27
74	Tay Sachs disease carrier screening in schools: Educational alternatives and cheekbrush sampling. Genetics in Medicine, 2005, 7, 626-632.	2.4	33
75	Educating general practitioners about prenatal testing: approaches and challenges. Prenatal Diagnosis, 2005, 25, 592-601.	2.3	14
76	A Long-term Outcome Study of Intersex Conditions. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 555-67.	0.9	139
77	Integrating genetics as practices of primary care. Social Science and Medicine, 2004, 59, 223-233.	3.8	40
78	Multimedia messages in genetics: Design, development, and evaluation of a computer-based instructional resource for secondary school students in a Tay Sachs disease carrier screening program. Genetics in Medicine, 2004, 6, 226-231.	2.4	15
79	Experiences at the time of diagnosis of parents who have a child with a bone dysplasia resulting in short stature. American Journal of Medical Genetics Part A, 2003, 122A, 100-107.	2.4	22
80	Needs assessment study of genetics education for general practitioners in Australia. Genetics in Medicine, 2002, 4, 71-77.	2.4	89
81	Demonstration of calcitonin gene-related peptide receptors in the gubernaculum by computerized densitometry. Journal of Pediatric Surgery, 1992, 27, 876-878.	1.6	37
82	Serum levels of mullerian inhibiting substance in boys with cryptorchidism. Journal of Pediatric Surgery, 1991, 26, 621-623.	1.6	54
83	Serum Levels of Mullerian Inhibiting Substance in Boys from Birth to 18 Years, as Determined by Enzyme Immunoassay*. Journal of Clinical Endocrinology and Metabolism, 1990, 70, 11-15.	3.6	93
84	Studies on species cross-reactivity of hemopexin by use of monoclonal and polyclonal antibodies. Biochemical and Biophysical Research Communications, 1987, 144, 88-93.	2.1	4
85	An avian serum .alpha.1-glycoprotein, hemopexin, differing significantly in both amino acid and carbohydrate composition from mammalian (.beta.-glycoprotein) counterparts. Biochemistry, 1986, 25, 6555-6562.	2.5	24
86	Possible mechanism for differences in sensitivity to cis-platinum in human prostate tumor cell lines. Cancer Letters, 1986, 31, 163-169.	7.2	31
87	Identification of synergistic combinations of spirogermanium with 5-fluorouracil or cisplatin using a range of human tumour cell lines in vitro. Investigational New Drugs, 1984, 2, 29-33.	2.6	16