



## **The NSGC Genetic Counselor Research Repository Compendia**

Following the development of the NSGC Genetic Counselor Research Repository (formerly the NSGC Bibliography), the Payer Subcommittee has created compendia (collections of concise but detailed information about a particular subject) to highlight key articles that are important in various topics related to the value of genetic counselors. These topics include:

- Arguing the Business Case: Economic Value of Genetic Counselors
- Improving Patient Care: Clinical Value of Genetic Counselors
- Increasing Access to Genetic Services and Growing the Genetic Counseling Workforce
- Preventing Harm: Gaps in Patient and Non-Genetics Provider's Knowledge of Genetics
- Promoting Efficiency and Quality of Genetic Service Delivery

Key articles are identified through a search of the Genetic Counselor Research Repository using relevant key words and consultation with individuals who are content experts in each particular topic. Each article is reviewed by members of the Payer Subcommittee for relevance and importance for inclusion in the compendium.

### **Preventing Harm: Gaps in Patient and Non-Genetics Provider's Knowledge of Genetics**

This collection includes key articles that demonstrate that there is need for genetic counselors in the healthcare workforce, that non-genetics providers would benefit from support in genetic care, and that genetic counselor involvement in care can prevent harm.

Baldwin LM, Trivers KF, Andrilla CH, Matthews B, Miller JW, Lishner DM, Goff BA. **Accuracy of ovarian and colon cancer risk assessments by U.S. physicians.** J Gen Intern Med. 2014;29(5):741-749. PMID: 24519100.

Survey of 1,555 physicians demonstrates misestimation of patient ovarian and colon cancer risk, with participants reporting both overestimations and underestimations compared to national guidelines.

Gilstrop Thompson M, Corsetti S, Jain V, Ruhstaller K, Sciscione A. **Accuracy of Routine Prenatal Genetic Screening in Patients Referred for Genetic Counseling.** Am J Perinatol. 2020;37(3):271-276. PMID: 30795016.

Determines the rate of missed genetic information in the general obstetrician's routine prenatal genetic screening process by comparing findings of a targeted genetic history form and genetic counseling appointment to genetic history obtained by the obstetrician provider.

Farmer MB, Bonadies DC, Mahon SM, Baker MJ, Ghate SM, Munro C, Nagaraj CB, Besser AG, Bui K, Csuy CM, Kirkpatrick B, McCarty AJ, Weiss McQuaid S, Sebastian J, Sternen DL, Walsh LK, Matloff ET. **Adverse Events in Genetic Testing: The Fourth Case Series.** Cancer J. 2019;25(4):231-236. PMID: 31335384.

Fourth article in case series. Documents 25 cases from cancer, pediatric, preconception, and general adult settings in which tests were recommended, ordered, inputted, or used incorrectly. The article also includes a discussion of strategies to increase access to genetic counselors.

Predham S, Hamilton S, Elliot AM, Gibson WT. **Case Report: Direct Access Genetic Testing and A False-Positive Result For Long QT Syndrome.** J Genet Couns. 2016 Feb;25(1):25-31. PMID: 26318596.

Case report of an individual who pursued genetic testing through direct-to-consumer methods: the individual received a positive result that ultimately turned out to be a false positive when confirmed with a clinical grade lab. The report explores the cost to the healthcare system, patient angst, and discusses the value of genetic counselors as a way to avoid the scenario in the future.

Brierley K, Campfield D, Ducaine W, Dohany L, Donenberg T, Shannon K, Schwartz R, Matloff E. **Errors in delivery of cancer genetics services: implications for practice.** Conn Med. 2010 Aug;74(7):413-23. PMID: 20806621

Analyzes 21 cases of adverse outcomes of cancer genetic counseling and testing performed by non-genetics providers. Major patterns included wrong genetic test ordered (for example, ordering BRCA1/2 testing instead of Lynch syndrome genes), misinterpretation of genetic testing results, and inappropriate, inadequate or lack of genetic counseling. Adverse outcomes included unnecessary health expenditures, incorrect medical management recommendations (including unnecessary surgeries and false reassurances), and psychosocial distress.

So PL, Cheng YKY, Cheuk KY, Chiu WK, Mak SL, Mok SL, Lo TK, Yung WK, Lo FM, Chung BHY, Kan ASY, Lee CP, Tang MHY. **Health professionals' involvement and information provision in genetic counseling following prenatal diagnosis of sex chromosome aneuploidy in Hong Kong.** Int J Gynaecol Obstet. 2019 Mar;144(3):314-316. PMID: 30516269.

Retrospective analysis of counseling records for pregnancies with prenatal diagnoses of sex chromosome aneuploidies. Compared to obstetricians, pediatricians and clinical geneticists were more likely to provide information about the cause, recurrence risk, and management of sex chromosome aneuploidies. They note that termination rate declined throughout the years, which the authors speculate may be due to increasing involvement of genetic specialists.

Skotko B. **Prenatally diagnosed Down syndrome: mothers who continued their pregnancies evaluate their health care providers.** Am J Obstet Gynecol. 2004 Nov;192:670-7. PMID: 15746657.

Survey of mothers of children with Down syndrome about their experience receiving a prenatal diagnosis of Down syndrome. A majority of respondents expressed frustration with the process, including lack of information before screening or diagnostic testing.

Vadaparampil ST, Scherr CL, Cragun D, Malo TL, Pal T. **Pre-test genetic counseling services for hereditary breast and ovarian cancer delivered by non-genetics professionals in the state of Florida.** Clin Genet. 2015 May;87(5):473-7. PMID: 24735105.

Survey of non-genetics professionals about their clinical practices offering *BRCA* testing. Participant responses demonstrated lack of adherence to guideline-based practice: few constructed a three-generation pedigree, discussed alternative hereditary cancer syndromes, or the meaning of a variant result.