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 demonstrates 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.


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.


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.


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.

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.


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.


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.


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.


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.