Genes & Diseases (2018) xx, 1–5

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: http://ees.elsevier.com/gendis/default.asp

REVIEW ARTICLE

The importance of preventative medicine in conjunction with modern day genetic studies

Sierra Sandler, Lauren Alfino, Mir Saleem*

NOVA Southeastern University, United States

Received 10 February 2018; accepted 8 April 2018

KEYWORDS
Cancer screening;
DNA testing;
Genetic abnormalities;
Genetic analysis;
Gene mutation;
Hereditary disorders;
Prevention of genetic diseases;
Primary care

Abstract Genetic screening in the primary care setting is the future of preventative medicine. Genetic testing is an important medical tool for assessing various inheritable diseases, conditions, and cancers. The ability to diagnose patients before symptoms surface can help lessen the severity of symptoms and promote quality of life. However, genetic screening can cause psychological distress from the knowledge of test results, in some cases only serving to increase the risk of developing a condition due to stress. Genetic testing can be conducted anytime in life, even before birth. In this review, a compilation of genetic testing's definitions and boundaries, factors influencing an individual's test outcomes, and an overview of a wide variety of diseases, conditions and cancers were collected.

Introduction

In the world of modern health care, innovation leads the way to not only better patient care, but also a deeper understanding of preventative medicine. The ability to treat a genetic disease before it may become severe is a scientific feat that, until recent years, has been unknown to physicians. Genetic testing saves countless lives that would otherwise fall prey to congenital diseases (i.e. Tay Sachs and Cystic Fibrosis) and cancers (i.e. breast and colon).[1–3] By understanding at a genetic level how a patient may be predisposed to certain health conditions, the planning for their care can start before an unfavorable prognosis, and

* Corresponding author. Dept. of Biology, Halmos College of Natural Sciences and Oceanography, NOVA Southeastern University, 3rd Floor, Parker Building, 3301 College Avenue, Fort Lauderdale, FL 33314, United States.
E-mail addresses: ss3287@mynsu.nova.edu (S. Sandler), la863@mynsu.nova.edu (L. Alfino), msaleem@nova.edu (M. Saleem).
Peer review under responsibility of Chongqing Medical University.

https://doi.org/10.1016/j.gendis.2018.04.002

Please cite this article in press as: Sandler S, et al., The importance of preventative medicine in conjunction with modern day genetic studies, Genes & Diseases (2018), https://doi.org/10.1016/j.gendis.2018.04.002

not hastily chasing after one.[4] Primary care physicians possessing the advantage of an early start on planned treatment gives us the ability to elude health issues that may later consume a patient's life. As a result, this surge in genetic screening in primary care physician offices can change lives.[2]

Unfortunately, for many diseases today genetic analysis will not be able to cure the patient completely ahead of the racing clock. However, it may have the ability to soften the blow of a debilitating disease by lessening the severity of symptoms or preventing it from manifesting early in life.[4] A physician's mission to care, cure, and comfort is enhanced with genetic screening. The faculty to understand and treat preventively can massively enhance the quality of life for a patient. In short, genetic testing in a primary care setting can transform otherwise sick lives into healthier, or even healthy lives.[4]

Genetic disorders

Genetic testing works by analyzing changes in DNA that have been linked to certain diseases, conditions and cancers.[5–8] There are over 7000 disorders that are believed to be linked to Mendelian genetics, and over 700 tests available currently.[9] Analyzing these changes through genetic testing is most useful when an individual has had family members affected by the condition, or if the disease could be linked to inheritance through the process of Mendelian genetics.[10,11] By seeing patterns of diagnosis in relatives, primary care physicians can use those past cases to recommend specific genetic tests to their patients.[5] Different diseases can be inherited in a variety of ways depending on the magnitude of influence the gene (or genes) being investigated may have.[4] If it is recessive or dominant, or if the severity is dependent on being heterozygous or homozygous in nature, this may also impact the severity of the condition.[6,12,13] More often than not, the severity of a disease or the probability of manifesting a disease's detrimental symptoms is increased when familial cases are common.[4] Genetic testing can also identify those that have a higher chance of showing symptoms of the condition or those that are at risk of passing it on if they have progeny.[6,7,10,12,14] This allows for primary care physicians to collect patient data for future generations, aiding in the tracking and prediction of future diagnoses.[15]

Various unavoidable genetic diseases that impact an individual from birth are critical to identify early in a patient's life. This is in order to make informed decisions about choices of planned care to improve quality of life.[6,7,12] Inherited genetic diseases of a recessive nature have been shown to account for nearly 20% of total infant mortality and 10% of infant hospitalization in the United States.[12] Other conditions, such as congenital deafness, can also be linked to hereditary causes.[7]

Down syndrome

The age of a mother has also been shown to have a negative impact on the occurrence of genetic diseases such as Down syndrome, with higher incidence development of negative reactions to drugs found in women who give birth older than 35 years of age.[16] Due to rising popularity in screening, older mothers can obtain a better understanding of how probable passing on a hereditary condition such as Down syndrome would be.[16,17] Screening for theses genetic diseases from preconception has been shown to greatly reduce these statistics and reduce mortality in infant cases stemming from these genetic conditions.[12,13]

Cystic Fibrosis, Tay Sachs, familial dysautonomia, and BRCA genes

Multiple factors influence whether an individual will be tested, two of which include considerations such as family history and ethnicity.[14] As stated before, genetic testing is best utilized when closely related individuals to the patient have tested positive for the mutation or have the condition.[10,14] Certain ethnicities are more likely to carry certain genes. For example, due to the knowledge obtained from genetic screening, individuals of Ashkenazi Jewish descent have a higher percentage of particular genetic disorders including as Cystic Fibrosis, Tay Sachs, familial dysautonomia, and the development of BRCA genes.[1,6,11,14,16] Thus, as part of primary care a physician may recommend screening due to a patient's background.[4] This enables the physician to preventatively identify conditions before they may have the ability to become debilitating. Genetic screening has seen a surge in popularity among demographics such as individuals of Ashkenazi Jewish descent, and this increase in screening has shown to decrease the number of Tay Sachs cases in said population.[11,14] With genetics becoming underlined as a point of study in modern primary care, other diseases could see similar patterns.[11,18] This would result in early diagnosis, as well as the disease prevalence in its respective population decreasing in a similar fashion to Tay Sachs. This means that looking forward, genetic testing can be applied to any disease's preventative treatment so long as the disease has proven to be hereditary.

Tay Sachs in the Ashkenazi Jewish population has greatly diminished from testing as well.[1,6] Early diagnosis of conditions such as Tay Sachs has been proven to favorably impact the survival and quality of life for those patients.[8,19]

Cancer

Colorectal cancer

One of the more understood hereditary cancers is colorectal cancer.[20] Several diversified genes have been linked to different cancers of the colon such as familial adenomatous polyposis (FAP), hereditary nonpolyposis colorectal cancer (HNPCC), and Turcot's syndrome which has been linked to the same genes being altered from FAP and HNPCC.[21] Mutations, or altering of the gene(s) involved, occurs in the germ lining which can subsequently lead to the development of cancer.[3,8,14,21] Testing positive for FAP, HNPCC or Turcot's syndrome does not guarantee an individual will develop cancer.[20] However, it does mean that an individual will have an increased likelihood of developing cancer and are without a doubt carrying the gene for its existence.[21] Thus, they have the ability to pass it on.[21] As with many cancers, testing positive for these different gene mutations increases the chance of developing the cancer.[20]

Please cite this article in press as: Sandler S, et al., The importance of preventative medicine in conjunction with modern day genetic studies, Genes & Diseases (2018), https://doi.org/10.1016/j.gendis.2018.04.002