Importance of Genetic Testing for Cancer Genes Association

Richa Sharma, Pooja Paliwal, Yogita Gigras

Abstract— Now a day cancer is a well known human disease which has approximately 200 different research types mutation cancer. Hereditary probs are preventing when mutation accurse in the genes of cancer cells and genes passed along generation to generation. The objective of this research work is to help in detecting genetic cancer and to enhance the performance of genetic testing. To verify and improved the effectiveness of genetic testing, it is test on breast and ovarian cancer. According to some research mutation is most common in people of eastern European (ashkenazi) jewish descent. Genetic testing used to determine changes in genes that that make to develop diseases like cancer. Soft computing method genetic testing used to classify the presence of breast and ovarian cancer.

Keywords—Hereditary, Mutation, Genetic Teseting, Genes.

I. INTRODUCTION

Hereditary of cancer can be different types of cancers like colon and uterine cancer, thyroid cancer, pancreatic and brain tumor and breast or ovarian cancer, these cancers formed when abnormal change in genes is called mutation. Those abnormal genes sometime passed generation to generation that inherited from parents is referred to as hereditary cancer. In humans, the child is formed by sperm (egg) and mutation occurs in child by inherited gene. An egg is fertilized by sperm and egg formed a cell, that's name a zygote which split to generate a fetus. Mutation in each cell in the body may be take place on the generation to generation. Because of this nature of hereditary cancer we perform genetic testing in which taking sample of blood, check tissue in order to analyze a person's genes. These tests measures diseases like cancer.

Now we discuss about one of the most common genetic cancer hereditary breast or ovarian cancer (HBOC) where BRCA1 and BRCA2 are the most common genes. For that cancer, check blood sample that indicates whether a person carries a changes in these genes which can increase cancer risk. HOBC can be occurred when mutation performed in either BRCA1 or BRCA2. Mutation in either BACA1 or BRCA2 gene can make breast or ovarian cancer risk very tremendous, but tend to be higher with BRCA1 mutation.

According to some survey risk of breast cancer 50% to 85% in lifetime and after age 50 it become 30% to 50%. For ovarian cancer mutation of BRCA1 gene is 25% - 50% and mutation of BRCA2 gene is 15% - 30%.

For some patients and families, genetic services area unit consultants in managing genetic condition of families, often over many generations, and providing each varieties of services clinical and laborat*ory* for any body system.

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The bulk of specialist says; genetic services or a 'genetic disorder', within the individual's gene sequencing different, may well be discovered in an exceedingly Medical Genetic Testing (MGT) Laboratory and clinics which offer through 'Regional Genetic Centers'. MGT science laboratory analysts would concern a MGT article once learning the gene alternative, with alternative depiction and explanation; then deliver it to the analysis appealing *clinicians*. At the time of this method, genetic science lab is massively promoted by info Systems / info Technologies (IS/IT) because

i) Information repository and data records, as an example, data processing and information tools; ii) Information analysis, e.g. the deoxyribonucleic acid (DNA) data Bank of Japan (DDBJ), Gen Bank and European molecular biological science Lab (EMBL) sequel database(DB), through base sequence referencing; adjustment search through Mutation Surveyor TM; phenotype- genotype parallel search through 'UMD (Universal Mutation DB) Central Phenotype-Genotype Analysis'; mutation search through 'UMD Central Mutation search'; iii) Info looking, e.g. data processing tools and National Center for Biotechnology dat*a* (NCBI) program, UMD Central tool, Google program etc.; and iv) Info dispatching, such as Email, Internet and so on.

II. HEREDITARY CANCER

Cancer could be a quite common illness for currently days, several family have a minimum of one or 2 members who have had cancer. everybody comprehend the causes of cancer like smoking and plenty of additional. however associate degree in a very variety of cases the cancer is caused through an abnormal sequence with the aim of passed it from generation to generation.

Merely concerning five-hitter - 100% of every kind of cancers are as a result of genes defect (called mutation) genetic from the parent. This analysis work highlights those cancers. Genes the very small part of the DNA(deoxyribonucleic acid). Most of the genes area unit same altogether individuals however few of genes slightly completely different in individuals that build distinctive one and all. Genes hold the directions on a way to build the proteins for destroying injury cells and the way to stay the cells stable. They manage things like eye color, hair color and height. they will conjointly having bound likelihood of diseases, like cancer a more. human born with all the cell in which all the genes is exist from birth .chromosomes and genes constantly found in cells, completely particular types of cells using particular type of cells. For example, bone cells use a unique set of genes than muscle cells use. If the cell doesn't want gene it will turned off and not used. A used cell is activated or turned on by gene.

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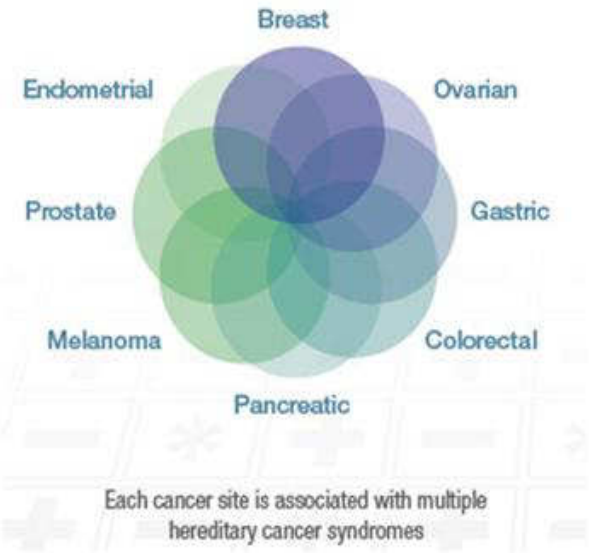
we have a pair of replica of genes – from each parent. Once somebody inherits genes abnormally copy then mutation already starts out in cells. If another gene's copy stop working then gene could be stop running generally. once the gene is stop functioning it will be a cancer perceptivity gene, It may develop cancer. Few cancer awaring genes will be work as tumor superior genes. Tumor superior genes are natural genes that reduce cell distribution that improve DNA mistake or inform cell when to depart. When tumor superior gene do not work accurately no of chances of cancer will increase if cell will increase disorderly. Most of the family cancer diagnostics are lead to by genetic defect of tumor superior gene.

Some individuals who are not birth with a nasty copy of a gene would need to achieve a pair of completely different mutations for that gene not to work. Receive a pair of mutations within the equivalent gene takes an expanded than starting one, so that cancers generate by genetic gene mutations likely appear in life than cancers of similar type aren't.

Pictorial view of hereditary cancer syndromes:-



III. Types Of Hereditary Cancer

Particular types of cancer can increase the risk factor of hereditary cancer because inherited gene faults present in their family members that is called family history of cancer. . These faulty genes will increment the probabilities of over one style of cancer like breast and ovarian cancer, colon and female internal reproductive organ (uterine) cancer and lots of additional. If you have family history of cancer it means you need genetic testing where genetic counselor check, how many number of cancer cases/chances in your family? Blow chart give the overview of some hereditary cancers:-

S. No.	Cancer	Syndrome	Gene	% of cancer in UK	Recommendation
1.	Breast and ovarian cancer	Breast-ovarian cancer syndrome 1 Breast-ovarian cancer syndrome 2	BRCA 1 gene BRCA 2 gene	3% Breast cancer and 2% ovarian cancer	Mammography and breast magnetic resonance imaging (MRI) and ovarian cancer screening for female family member.
2.	Hereditary colon cancer	Familial adenemetous polyposis	APC gene	6% in men and 5% in women	Removal of colon and life time cancer screening.
3.	Pancreatic and brain tumor	Familial melanoma	At least one of three genes	1 to 2%	Monthly skin self-exam and every six month examination by dermatagist.
4.	Colon and uterine cancer	Hareditary nonpoly posis colon cancer (HNPCC)	One of At least four genes	5%	Colon and uterine cancer screening at the beginning of young age and other screening based on family history.
5.	Thyroid cancer	Multiple endocrine neoplasia type 2A (HNPCC)	RET gene	5 to 20%	Removal of the thyroid grand.
6.	Kidney cancer	Von Hippel Lindau (VHL)	VHL gene	2%	Examination of the eye, kidneys, brain and spinal cord on yearly basis.

A. concerning breast cancer BRCA1 and BRCA2 genes

BRCA1 and BRCA2 are human genes that accrued risk of breast cancer, sex gland cancer and prostate cancer. once either of those genes is mutated, or altered, desoxyribonucleic acid harm might not be repaired properly and develop further genetic alterations which will result in cancer.

BRCA1 and BRCA2 growing the danger of feminine breast and sex gland cancers attributable to Unambiguous genetic mutations, and that they are accrued risks of many further varieties of cancer. BRCA1 and BRCA2 mutations criteria in percent:

1)Hereditary breast cancers account concerning 20 to 25 % of BRCA1 and BRCA2 mutations

2) concerning 5 to 10 % of all breast cancers and

3) BRCA1 and BRCA2 mutations in account for around 15 % of overall ovarian cancers. Breast and ovarian cancers related with BRCA1 and BRCA2 mutations likely growing at modern period of humans.

A dangerous BRCA1 or BRCA2 genes mutation is hereditary from a individual's parent. If kid of a parent carries a mutation in one in every of these two genes encompasses a fifty % risk of heritable the mutation. once a individual's second copy of the gene is traditional, the results of mutations in BRCA1 andBRCA2 are seen.

B. BRCA1 or BRCA2 mutation grow chances of woman's breast and female internal reproductive organ (ovarian) cancer

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If Women inherits a harmful BRCA1 or BRCA2 mutation, it extremely exaggerated chance of developing breast and/or female internal reproductive organ cancer.

Breast cancer: within the general population approx 12 % of ladies can develop breast cancer someday throughout their lives. per the recent estimates, approx fifty five to 65 % of ladies who succeed a dangerous BRCA2 mutation. BRCA2 mutation can establish breast cancer after the age of 70 or above in women.

Ovarian cancer: within the general population approx 1.3 % of ladies can develop female internal reproductive organ cancer someday throughout their lives. per the recent estimates, approx 39 % of ladies who genetically damaging BRCA1 mutation and approx 11 to 17 % of ladies who inherit a damaging BRCA2 mutation.BRCA2 mutation can establish female internal reproductive organ cancer by age seventy years.

Additional characteristics of a woman that have cancer risk, will produce her risk higher or below the common, it's terribly important note for everybody. case history of a selected girl comprise these characteristics of breast, female internal reproductive organ and alternative cancer possibility; the particular mutation she has genetically; and alternative factors, like her generative history.

B. More cancers that are raised mutations in BRCA1 and BRCA2

Several cancer in addition to breast and female internal reproductive organ cancer that increase the chance of harmful mutations in BRCA1 and BRCA2. Because of BRCA1 mutations uterine tube cancer and peritoneal cancer could increase the chance of developing these cancers. Men by means that of lesser quantity BRCA1 mutation and BRCA2 mutation are exaggerated risk of carcinoma. Men through adverse BRCA1 or BRCA2 mutations have the next chance of prostate cancer. BRCA1 or BRCA2 mutations could also be at exaggerated risk of pancreatic cancer in each Men and ladies. Mutation in BRCA2 is additionally referred to as FANCD1, if they're familial from each folks, will cause a Fanconi anemia subtype (FA-D1), a syndrome that's related to childhood solid tumors and development of acute myelocytic leukemia. Similarly, a mutation in BRCA1 is additionally referred to as FANCS, if they're familial from each folks, will cause another Fanconi anemia subtype.

D. Mutations in BRCA1 and BRCA2 further common in definite racial/ethnic populations than others

As during this research work beginning offer associate degree example, individuals of ashkenazi someone descent have the next prevalence of harmful BRCA1 and BRCA2 mutations as comparison to individuals within the general U.S. population. round the world alternative ethnic and geographic populations, for instance the Norwegian, Dutch, and Icelandic peoples, further have the next prevalence of precise harmful BRCA1 and BRCA2 mutations. Inadequate knowledge further indicate that the prevalence of specific harmful BRCA1 and BRCA2 mutations could also be totally different among individual racial and ethnic teams within the us, together with African Americans, Hispanics, Asian Americans, and non-Hispanic whites.

IV. GENETIC TESTING

genetic testing is the testing of genes, chromosomes or proteins that take a look at verify the management (KM) technology deficiency in genetics domain, then proposes an information system (IS) design to determine method automation and content management of the appropriated advancement of knowledge generation and knowledge management (KG& amp; KM) throughout MGT result interpretation. The given IS can validate the interpretation call by exploitation information Systems / data Technologies (ISIIT), esp. KM tools, like work flow Management System (WFMS), search engine and software package. Once developed and enforced, our integrated system can considerably improve MGT science laboratory researchers' KG& KM performance through increasing information capture, rising documentation quality and maintaining (if not improving) users' data satisfaction.

sorts of genetic testing

- Newborn screening: simply when birth newborn screening are often performed and accustomed acknowledge genetic disorders.
- Diagnostic testing: diagnostic testing is employed to spot before birth or any time in throughout the individuals life. once specific condition is suspected like physical signs and symptoms.
- Carrier testing: Classify people that carry one copy of point mutation that, once gift in 2 copies, caused of genetic disease. If each folks square measure tested, the take a look at will offer data a few couple's risk of getting kid with a genetic condition.
- prenatal testing: prenatal testing are often accustomed acknowledge changes in an exceedingly vertebrate genes before birth throughout physiological condition. It will noted conclude all attainable genetic disorders and birth defects.
- Preimplantation testing: it's accustomed notice changes in embryos that were created exploitation power-assisted fruitful techniques like in-vitro-fertilization. this system will reduse the chance of getting kid.
- Predictive and diagnosis testing: this sort of testing are often accustomed notice factor mutations related to disorders that seem when birth. These take a look at will useful to individuals United Nations agency's loved one have genetic disease however who don't have any options of the time of testing the disorder themselves.
- Presymptomatisting: will verify whether or not someone can develop a genetic disease like hereditary homochramatosis previous mark or warning The results of these testing will offer data a few person risk of developing a particular disorder and facilitate with creating selections concerning treatment. it use deoxyribonucleic acid Sequences to spot a private for legal purpose.

Who ought to contemplate genetic testing for BRCA1 and BRCA2 mutations?

Harmful mutation in BRCA1 and BRCA2 gene are relatively rare within the common population,

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most specialist agree that mutation testing of people ought to be performed only the person's case history recommends the probable presence of a harmful BRCA1 or BRCA2 genetic mutation in individuals who don't have cancer. Women who have members of the family with breast, ovarian, uterine tube, or peritoneal cancer evaluated, consistent with the survey in Dec 2013 of the united states Preventive Services Task Force.

By using transmission equipment we will provide health care analysis . These equipment measures ancestors history of harmful BRCA1 and BRCA2 genetic mutation factors, including:

1. Cancer in each breasts within the same lady
2. Breast cancer diagnosed before age fifty years
3. breast and ovarian cancer will be found also similar women or in a ancestors.

Several breast cancers

4. Two or a lot of primary forms of BRCA1 or BRCA2 connected cancers in a single family member

Some cancer cases of male breast.

People of ashkenazi jewish ethnicity:

Once a personal contains ancestors' history of the presence of BRCA1 or BRCA2 mutation, initial check a ancestors member

If in the family their is a member who is cancer patient and remains full of like and, it's going to be most informative for alternative members of the family counsel. Counsel is important for alternative members of the family to find out their ability chance and whether or not heritable testing for mutations in BRCA1 and BRCA2 can be applicable for them.

If it is unfeasible to verify the occurence of a risky BRCA1 or BRCA2 mutation in ancestors those who have cancer, it's applicable for each man and ladies who don't have cancer however have a relative anamnesis that means the occurenceof such a mutation to own counsel for potential testing.

Several people—for example, people who were adopted at birth—may not recognize their case history. In cases wherever a lady with an unknown case history has an early-onset breast cancer or female internal reproductive organ cancer or a person with an unknown case history is diagnos with breast cancer, it's going to be affordable for that people to contemplate genetic testing for BRCA1 or BRCA2 mutation. people with an new case history who don't have an early cancer or male breast cancer are at terribly low risk of getting a harmful BRCA1 orBRCA2 mutation and are doubtful to profit from regular heritable testing.

Professional societies don't advocate that kids, even those with a case history implicational a damaging BRCA1 or BRCA2 mutation, bear heritable testing for BRCA1 orBRCA2. this can be as a result of no risk-reduction methods exist for kids, and children's risks of developing a cancer kind related to a BRCA1 or BRCA2 mutation are extraordinarily low. when kids with a case history implicational a harmful BRCA1 or BRCA2 mutation become adults, however, they'll need to get counsel regarding whether or not to undergoing genetic testing.

Is folks consider genetic testing for BARCA1 and BARC2 mutation speak with a heritable counselor?

Genetic counseling is mostly suggested earlier than and when any heritable analysis for an hereditary cancer syndrome. This counselling ought to be performed by a health care skilled who is intimate in cancer heritable. Counsel sometimes covers several feature of the testing method, as well as:

A genetic cancer chance assessment review entity's personal and family medical record Discussion of:

- The suitableness of heritable testing
- Positive and negative medical implications of a checked answers.
- The chances of checked result should not be helpful.
- The psychological chance and edges of genetic check results
- The risk of passing a mutation to kids

Discription of the precise test which may be used and also the precise Risk factors that increases and decreases a women's risk of developing cancer

- Family history: when close relatives like mother-father and some other maternal-paternal relatives have had particular cancer is called family history. This factor increased the risk because of an inherited faulty gene.
- Genetic mutation: gene mutation can change in sequences of DNA that increase the chance of developing cancer. genetic mutation or gene mutation is two varieties somatic and hereditary mutation. Hereditary mutation is inhibit in parent's sperm cell and inherited kid that grows from the fertilized egg. somatic mutation can be accrue by environmental feature like ultraviolet rays from sun or if a blunder accrue in DNA copies itself throughout cellular division.
- Harmon replacement: Harmon replacement therapy is risk factor of cancer.
- Increased age: Increasing age is one of the main risk factor of cancer because of genetic changes accrue in body as well as age increase.
- More menstrual cycle.

A. ***Decreasing risk factors of cancer in women***

Removal of ovaries/fallopian
Childbearing
Breastfeeding
Tubal ligation
Oral contraceptive use.

V. Conclusion

This work described the different types of hereditary cancer and about genetic testing, there are many avenues for future research. First uniquely identify the genetic testing of hereditary cancer than investigate the better and more useful way of genetic testing by which improvements come in hereditary cancer diagnosis system.

FUTURE WORK

Now day genetic diseases and genes affiliated with them is a common problem of human health. Many algorithms used for optimal gene subset selection by which easy to

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classify genetic diseases and its related genes. In this research work we apply one of them for a particular cancer disease. And select best section method by comparing more than one method.

REFERENCES

1. Yulong Gul, James Warren2, Jan Stanek', Graeme Suthers3“A System Architecture Design for Knowledge Management (KM) in Medical Genetic Testing (MGT) Laboratories” University of South Australia, Australia.
2. Matthews, Dr. Anne. "Prenatal Genetic Testing." Expectant Mothers Guide. Spindle Publishing Company, n.d. Web. 17 Dec. 2010.
3. Kaplan, Karen. “Genetic Tests Offer Knowledge, Not Wisdom.” Los Angeles Times (Los Angeles, CA). Sept. 19 2007: n.p. SIRS Researcher. Web. 15 Dec 2010.
4. http://Ghr.nlm.nih.gov/handbook/testing/uses
5. http://www.cancer.net/cancer-types/hereditary-breast-and-ovarian-cancer
6. http://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet
7. http://www.cancerresearchuk.org/about-cancer/causes-of-cancer/inherited-caancer-genes-and-increased-cancer-risk/inherited-genes-and-cancer-types
8. http://www.siteman.wustl.edu/ContentPage.aspx?id=4512

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