Genetics in Medicine

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Advances in Genetics are transforming medicine faster than ever before. Genetics is everywhere in medicine. This article briefly reviews how genetics is making an impact in medicine.

Genetic testing is used to diagnose inherited diseases. Take the example of thalassaemia. There are two types of thalassaemia – alpha thalassaemia and beta thalassaemia. When a child is diagnosed to have thalassaemia a genetic test can be used to find out whether the mutation is in the alpha gene or the beta gene. If the mutation is in the alpha gene, then the child has alpha thalassaemia. If the mutation is in the beta gene, then the child has beta thalassaemia. Genetic testing is also useful to diagnose acquired diseases that are the result of a genetic change. In case of these diseases, the person gets normal genes from his/her parents, but he/she develops a genetic change in his/her own cells resulting in an illness. There are many examples of this. One of the well known examples is chronic myeloid leukaemia (CML). In this condition, some cells in the bone marrow acquire a chromosomal abnormality – a balanced translocation between chromosome no. 9 and chromosome no. 22. This translocation results in the creation of an abnormal fusion gene called the bcr/abl fusion gene. This is transcribed in to the bcr/abl mRNA. The abnormal mRNA is translated into a fusion protein which causes CML. We can diagnose CML by detecting the presence of bcr/abl mRNA in the body of a person who is suspected to have the condition based on bone marrow histopathological findings using a genetic test. Not only that, once the person is treated, we can find out whether he/she has responded to treatment by finding out whether the abnormal cells have cleared from the body by repeating the same test. These are only two examples of genetic diagnostics. Thousands of conditions can now be diagnosed based on genetic tests. You can read about them at http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests.

Genetic testing can also be used to detect infectious diseases. There are many examples. The common ones are the genetic tests for Mycobacterium Tuberculosis, Hepatitis B virus, Hepatitis C virus, cytomegalovirus, and Dengue virus. These enable fast and reliable diagnosis of infectious diseases. Take for example the genetic test for the Dengue virus. It is very commonly used in people with fever to find out whether they have Dengue. The test for cytomegalovirus is used commonly in a special

group of people, people who have undergone kidney transplantation. These people are prone to cytomegalovirus infection. Antiviral treatment for cytomegalovirus is very expensive. So finding whether the patient has the infection definitely is important. In addition it is necessary to find out whether the patient has responded to treatment by repeating the test after treatment. A special genetic test called quantitative real time PCR makes it possible to do that. It is the same type of test that is used in the case of the bcr/abl mRNA that was mentioned before.

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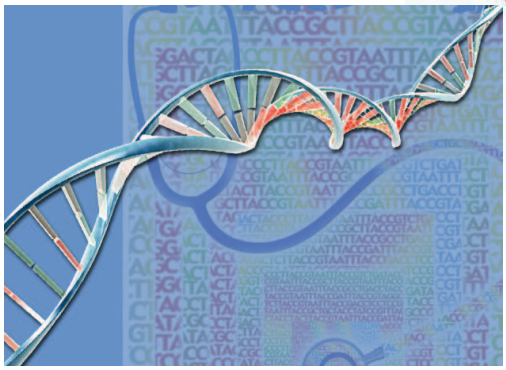
People are protected against infectious diseases by immunizing using vaccines. Recombinant genetic technologies are used to develop vaccines against infectious diseases. Most vaccines now in the market are based on recombinant genetic technology. Infectious agents however, undergo change and develop resistance to commonly used antibiotics. Genetic testing can be used to identify resistant strains of bacteria. In addition knowledge about the structure of bacterial as well as viral pathogens can be used to design new antibiotics to combat infectious diseases. Genetic knowledge is also used to develop treatment for other illnesses. Again take the example of CML. The treatment for CML is a drug that blocks the action of the bcr/abl fusion protein. Recombinant gene technology can also be used to make therapeutic products (for example, insulin, interferons) more affordable.



Medicine is not all about diagnosing and treating illnesses. It is also about creating an environment in which people can live a healthy life. Genetic technologies are used to improve the environment – e.g. sanitation, clean water, bioremediation, etc. In addition genetically modified food with increased nutrients have been developed to counter nutritional deficiencies

Genetics was once thought of as most useful for those who could pay for it in affluent countries. The appreciation that much could be done for the developing world about a decade ago (1,2) resulted in a shift in emphasis which has made it possible for us in the developing world to benefit from these technologies. Genetics is not a foreign concept anymore. Genetics is here to stay in Sri Lanka. Hundreds of genetic tests are ordered by doctors in Sri Lanka daily. The people of this country are increasingly experiencing genomic medicine when they seek health care.

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