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Editorial

Prevention genetics program is an efficient model for precision medicine

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Precision medicine also called personalized medicine or individualized medicine has become a major focus of health systems over the world. It is defined by National Institute of Health as a novel treatment and prevention method based on the understanding of individual gene, environment, and lifestyle.[1,2] Genetic disorders could be one of the targets to apply the precision medicine techniques. Genetic disorders are quite prevalent in Saudi population specifically autosomal recessive disorders, and this is not surprising because of high rate of consanguinity.[3] There is no exact figure available on incidence of the inherited genetic disorders in Saudi Arabia; however, from the initial result of newborn screening program in Saudi Arabia for several disorders, the incidence was found to be 1:1000.[4] Despite intensive education and management, these genetic disorders are still propagating in Saudi population and their burden is worsening with the progression of time.[5] The most efficient mode of prevention is the primary prevention which proposes prevention of the disease before it ever occurs. This can be achieved for genetic disorders through several modalities such as perinatal genetic testing and preimplantation genetic diagnosis (PGD). Prenatal genetic testing is largely dependent on chorionic villus sampling (CVS) which is an invasive screening test that involves taking a small piece of tissue from the placenta. It is usually performed between the 10th and 12th weeks (1st trimester). CVS can be performed through the abdomen, which is called a transabdominal test, or through the cervix, which is called a transcervical test. Another type of sampling is called amniocentesis and is performed between 14th and 16th weeks (2nd trimester). During amniocentesis, samples from the amniotic fluid, which surrounds the fetus and contains fetal cells, are collected from the uterus using a needle. Both procedures carry a small risk of fetal loss.[6] The DNA is extracted from the samples and tested for specific gene mutation found in the family. If the result appears positive, then pregnancy may be terminated before the 19th week of gestation. The another modality is PGD which is a technique used to identify genetic defects in embryos created through *in vitro* fertilization before pregnancy. Both approaches show their efficiency and cost-effectiveness.

Genetics Division at the Department of Pediatrics, King Abdullah Specialized Children Hospital, Ministry of National Guard-Health Affairs, Riyadh, Saudi Arabia, started prevention genetics program by perinatal genetic testing method in January 2016. Since then, many cases have been tested and diagnosed prenatally, wherein 39% of the fetuses were confirmed to be affected with a known genetic disorder. Primary prevention was applied on 87% of the total fetuses. There is a significant increase in the volume of tested cases by 37% in 2017 compared to 2016. Therefore, the prevention rate has risen to approximately 60%. This program is a clear example of precision medicine as it involved customization of health care to prevent single genetic disorders discovered in a single family. It is cost-effective too as it appears to save 13,507,141.12 SAR annually. Such programs should be supported and funded to be a national program involving the whole Saudi Arabia rather than keeping it limited to a specific region. Prevention is the gold standard cost-effective and cost-saving measure to reduce the burden of genetic and metabolic diseases on the health-care system.

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