Hereditary breast and ovarian cancer

When healthy cells in the breast or ovaries change and grow out of control, they form a mass or sheet of cells called a tumor. A tumor can be malignant (cancerous) or benign (non-cancerous). A cancerous tumor grows and spread to other parts of the body whereas a benign tumor doesn't spread. Breast or ovarian cancer spreads when the cancer grows into other parts of the body or spreads (metastasis). Hereditary Breast and Ovarian Cancer (HBOC) is an inherited genetic condition where the cancer risk is passed from generation to generation in a family. BRCA1 and BRCA2 genes are associated with the majority of HBOC families. BRCA stands for BReast CAncer.

A mutation (alteration) in either BRCA1 or BRCA2 gives a woman an increased lifetime risk of developing breast and ovarian cancers and prostate cancer in men. Not all families with multiple cases of breast and ovarian cancer have mutations in BRCA1 or BRCA2. Normally, every cell has 2 copies of each gene: 1 inherited from the mother and 1 inherited from the father. Risk of getting HBOC increases even when mutation happens in only one copy of the gene for the person and such pattern is termed as autosomal dominant inheritance pattern. This means that a parent with a gene mutation may pass along a copy of the gene with the mutation to the child. Roughly a child has 50 % chance of inheriting the mutation from the parent with a mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having inherited the same mutation. About 10% to 30% of women under the age of 60 diagnosed with breast cancer, have BRCA1 or BRCA2 gene mutation.

HBOC is most frequently diagnosed when there are multiple cases of breast cancer and/or ovarian cancer on the same side of the family (approximately 80%). Effective screening methods have impacted on survival in cancer, but at present there is no effective screening test for ovarian and breast cancer based on gene patterns. Since diagnosis at an early stage is associated with improved rates of survival, the main objective of the proposed work is to diagnose HBOC earlier by identifying the alteration in the gene pattern that causes the abnormal phenotype or increases the disease risk. The diagnosis of BRCA1- and BRCA2-associated hereditary breast and ovarian cancer (HBOC) is proposed to be established in a person under study by identification of a heterozygous pathogenic variant in BRCA1 or BRCA2 using deep learning neural network (DLNN). Deep learning neural network is the best available technology for handling such vast and non-linear human gene based data. Hence we propose a DLNN based classification model for diagnosis of BRCA1 and BRCA2 associated HBOC earlier by identifying the alteration in the gene pattern that causes the abnormal phenotype or increases the disease risk. Such diagnosis would aid in increasing the survival rate of cancer victims and save millions of human life.