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Impact of copy number variation on common and complex diseases

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The ultimate goal of human genetics is to understand the role of genome variation in elucidating human traits and diseases. Although gene mapping strategies like genome-wide association studies (GWAS) had shown some success stories in the common and complex diseases, the inheritance of these diseases are largely unknown hence the "missing heritability" is unresolved. Besides single nucleotide polymorphism (SNP), copy number variation (CNV) defined as gains or losses of a DNA segment larger than 1 kb has recently emerged as an important tool in understanding heritable source of human genomic differences. It has been shown to contribute to genetic susceptibility of neuropsychiatric diseases, autoimmune diseases and some rarer and highly inherited cardiovascular diseases such as dilated cardiomyopathy. Using the infectious disease (Dengue) and cardiovascular disease (hypertension related left ventricular hypertrophy) as the models, we prove that the rare structural variants could contribute to the susceptibility of common and complex diseases. We revealed that the rare copy number variants altered the cell signaling related to immune response of the host therefore leading to vascular leakage in dengue. On a separate note, we also showed that the "fetal cardiac gene program" is altered by the rare copy number variant therefore resulted in left ventricular hypertrophy during pressure overload.

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Juvenile breast cancer: Presentation of a rare case

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Introduction: Younger women generally do not consider themselves to be at risk for breast cancer and in fact, just fewer than 7% of all breast cancer cases occur in women under 40 years old. The incidence is strongly associated with the appearance of the prognostic factors. Juvenile breast cancer seems to be more aggressive in comparison with other age-related breast cancer types. We present a case of a 23 year old female patient with appearance of multifocal breast cancer successfully diagnosed and treated.

Case: A 23 year old female patient was admitted to our Department complaining of pain and presence of a palpable mass located at her right breast. The physical examination confirmed this atypical appearance. The breast ultrasound and mammography revealed the presence of multifocality (hypodencic lesions at 10th, 11th hour and near the nipple). Due to the multifocality of the lesion a FNA of these areas was performed. The FNA examination confirmed the malignancy of the lesion. The preoperative staging of the lesion (bone scanning, CT thorax and abdominal CT) did not reveal any signs of metastatic infiltration. The patient underwent total right mastectomy. The sentinel node biopsy was positive for malignancy. After carrying out the mastectomy, a total axillary dissection was followed. (9/45 lymph nodes were infiltrated). The patient was discharged from the hospital in a good clinical condition on the 5 pod. Depending on the multidisciplinary decision, the patient is undergoing cycles of chemotherapy, hormonal therapy (ER+,PR+ receptors) and radiotherapy.

Discussion: Breast cancer is very rare in adolescents and very young women. Invasive breast cancer occurring in women before the age of 35 years has a more aggressive biological behaviour and is associated with a worse prognosis than in older premenopausal women. Breast cancers in these young women are more frequently poorly differentiated oestrogen-receptor (ER)-negative have lymphovascular invasion and high proliferating fractions. Breast conserving methods are accompanied with high recurrence rate and should be offered adjuvant therapy.

Conclusion: Juvenile breast cancer represents a rare entity in comparison with all other age-related types. It is characterized by the aggressive infiltration and the high recurrence rate. Multidisciplinary approach is mandatory in order to establish the ultimate confrontation.

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