ABSTRACT

The apparent association between schizophrenia and velocardiofacial syndrome (VCFS) has received attention lately in light of recent findings in linkage studies implicating chromosomal region 22q12, which is telomeric to the VCFS region, as a candidate region associated with genetic susceptibility to schizophrenia. VCFS, initially described in1978, is associated with a hemizygous deletion at chromosome 22q11. The present thesis aims to 1) review the various aspects velocardiofacial syndrome which is relevant to psychiatrists so as to improve the recognition of such syndrome 2) to describe two patients with schizophrenia and VCFS identified from the preliminary result of a pilot study of screening for deletion 22q11 in patients with schizophrenia in Hong Kong Chinese. The involved methodological limitations are discussed. The various clinical aspects of VCFS are reviewed with the emphasis of the wide range of phenotypic variability of the syndrome. The evidence for an increased risk of schizophrenia in VCFS is examined. The detailed case history of both patients is presented. In patient A, the significance of presence of cavum septum pellucidum is discussed in detail.