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Editorial

Schizophrenia, CATCH 22 and FISH

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Velo-cardio-facial syndrome (VCFS) is a syndrome of congenital abnormalities associated with small chromosomal deletions in the q11 band of chromosome 22. It was first reported by Strong in 1968 and further delineated by Shprintzen *et al* in 1978. Characteristic features include cleft lip and/or palate, hypernasal speech, **congenital** heart defects, learning disability and a **distinctive**ly long facial appearance with a bulbous nasal tip (Fig. 1). It has recently become apparent that VCFS is one of several overlapping phenotypes associated with deletions of 22q11 including DiGeorge syndrome, conotruncal anomaly face syndrome and sporadic or familial cardiac defects. The principal characteristics of these disorders have been summarised in the memorable acronym CATCH 22 representing *c*ardiac defects, *a*bnormal facies, *t*hymic hypoplasia, *c*left lip and/or palate, *h*ypocalcaemia and chromosome *22*q11 deletions (Wilson *et al*, 1993).

These disorders are of interest to psychiatrists because VCFS appears to have a characteristic behavioural phenotype including a bland affect, monotonous voice, impaired attention and poor social interaction with extremes of disinhibited or shy behaviour (Golding-Kushner *et al*, 1985). Of even greater interest is that, as cohorts of VCFS patients have been followed-up into adult life, a significantly increased incidence of psychosis has been observed. Shprintzen *et al* (1992) reported that more than 10% of their sample of greater than 100 patients with VCFS developed psychiatric disorders, mostly "chronic schizophrenia with paranoid delusions". More recently, Pulver *et al* (1994) also found a higher than expected rate (29%) of psychotic disorders (DSM–III–R schizophrenia or schizoaffective disorder; American Psychiatric Association, 1987) among VCFS patients with significantly higher rates of psychosis in second- and third-degree relatives.

This poses the interesting question of whether and to what extent 22q11 deletions are present in patients with schizophrenia.

Lindsay *et al* (1995) screened 100 unrelated patients and found chromosome 22q11 deletions in two patients. These results are interesting and provocative but clearly need replication and extension to larger samples.

This association with VCFS may have important implications for the aetiology of schizophrenia and suggests that 22q11 might contain a gene or genes of more general relevance to the disorder. It also suggests that clinicians should seek the clinical features of CATCH 22 in patients with schizophrenia or schizoaffective disorder. The presence of cleft lip and/or palate, characteristic dysmorph-

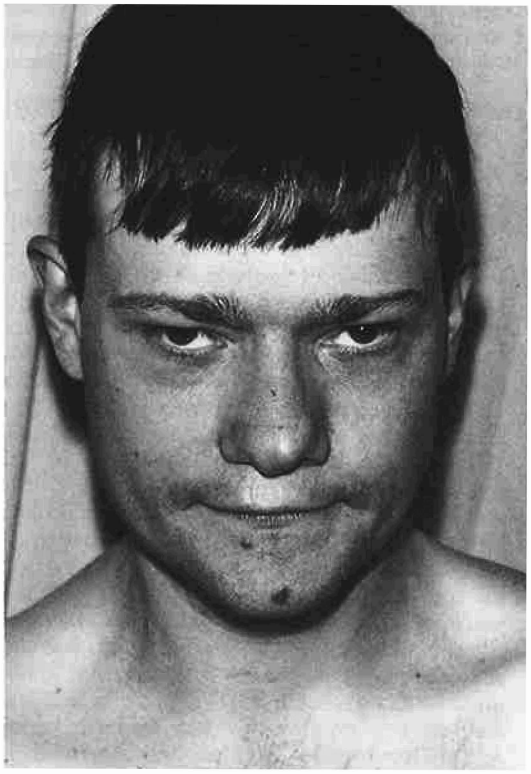

Fig. 1 Characteristic facial appearance in a patient with VCFS and DSM–III–R schizophrenia.

ology (Fig. 1), learning disability, congenital heart disease or hypocalcaemia should raise the suspicion of a chromosome 22q11 deletion. In such cases, psychiatrists should request molecular cytogenetic studies of chromosome 22q11 using fluorescence *in situ* hybridisation (FISH). In those patients demonstrating a deletion, close liaison with the local Department of Medical Genetics is recommended to ensure that appropriate genetic counselling is provided.

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