

($n = 23$). The chromosomal anomalies in these patients were mostly of *de novo* origin except in six cases (patients #12, #13, #16, #17, #18 and #19). In five cases parental chromosome study could not be performed (patients #9, #10, #11, #14 and #21). Marker chromosomes with an unknown origin found in three *de novo* cases. Sex chromosome aneuploidy was detected in six patients. Twelve cases had inversion 9q which is believed to be a normal variant.

DISCUSSION

There is great variation in the frequency of the reported chromosomal abnormalities found in MR patients. A cytogenetic study of 419 MR school children in southern Taiwan, by Shiue *et al* [2], found chromosomal abnormalities in 22.43% of the cases, with trisomy 21 occurring in 77 cases (18.38%). Sex chromosome aneuploidies were found in three cases (0.72%). Structural abnormalities of autosomes were found in 13 cases (3.10%) (2). Another study of 341 MR children in Taiwan found chromosomal abnormalities in 89 cases (20.3%) including 63 of trisomy 21 (10.7%) and 13 of fragile X (3.8%) [4].

Coco and Penchaszadeh [5] reported on a cytogenetic study in 200 MR children in Argentina. They found chromosomal abnormalities in 42.21% with 26 cases having structural chromosome defects [5].

Two studies were performed in The Netherlands. One study done in Amsterdam (in the south of The Netherlands) indicated that a chromosomal base in 22.1% of the patients was responsible for their MR. Of these, 14.3% were Down's syndrome patients, and 6.1% had other chromosomal abnormalities [6]. Another study done in Amsterdam indicated that 20 patients had chromosomal anomalies (7.5%) in 266 karyotyped MR children. Interestingly, these were mainly structural chromosome aberrations [7].

A study performed in Poland showed that the incidence of abnormal karyotypes in MR patients was 10.1% [8]. However, the percentage of chromosome aberrations found in patients with non specific mental retardation was 2.2% [8]. A study done by Butler and Singh [9] in America showed that 39 out of 201 (6.6%) institutionalized MR patients had abnormal chromosome with Down's syndrome noted in 31 of the patients.

While the overall frequency of chromosomal abnormalities in these reports was similar, there are reports of either low or high percentages of chromosomal aberrations in other studies. For example,

Celep *et al.* [10] reported the percentages of chromosomal abnormalities in 457 Turkish MR Patients to be only 4.81%. Chromosomal abnormalities and polymorphisms were detected in 65 (14.21%) (structural and numerical chromosomal abnormalities in 22 patients and polymorphisms in 43) of 457 MR and/or multiple congenital anomaly (MCA) patients. On the other hand, a study done in Slovakia revealed a very high percentage of chromosome abnormalities in MR patients. Of 324 MR patients, 104 (53.0%) had chromosomal aberrations [11].

The differences between the incidences of chromosomal abnormalities in the literature could be caused by the criteria for patient selection, and the techniques applied [cytogenetics only or in combination with molecular cytogenetics such as fluorescent *in situ* hybridization (FISH) and comparative genomic hybridization (CGH)]. In our study of 865 screened subjects, chromosomal anomalies were identified in 205 of the patients (23.6%). The majority were Down's syndrome cases ($n = 138$, 15.9%). Interestingly, we found three cases with marker chromosomes (0.34%). Licht and Weise [15] found that the incidence of marker chromosomes is about 0.288% in MR patients.

In general, as Karnebeek *et al.* [3] showed that the near yield of chromosome aberrations in classical cytogenetics is about 9.5% (variation: 5.4% in school populations to 13.3% in institute populations; 4.1% in borderline-mild MR to 13.3% in moderate-profound MR; more frequent structural anomalies in females). They also indicated that for fragile X anomalies, yields were 5.4% (cytogenetic studies) and 2.0% (molecular studies) [3].

The incidence of fragile X positive cases in our study is slightly higher than some other reports although we only employed cytogenetic tests for fragile X. For example, Butler and Singh [9] reported 2.0% fragile X positive in his cases, while in our study it was 3.8%. Nevertheless, our results indicate that the diagnostic contribution of the fragile X screening could be considered equally important as conventional chromosome banding techniques for the detection of structural chromosome abnormalities.

Some of the chromosome aberrations were detected in more than one case. For example: in two cases, chromosome 2 was involved with a very close breakpoint of q22 and q23 (Table 1; patients #1 and #14); in two cases, chromosome 4 with breakpoints p16 and p15.3 (Table 1; patients #5 and #15); and in