

# Fragile X syndrome. Clinical analysis of 300 Chilean patients with unspecific mental retardation Síndrome de X frágil. Análisis clínico en 300 pacientes con retardo mental inespecífico en la población chilena

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Background: Fragile X syndrome is the most important cause of sex linked mental retardation and the second of chromosomal origin, after Down syndrome. Aim: To apply the modified Hagerman score to patients with mental retardation and to relate clinical findings with cytogenetic and molecular diagnosis. Patients and methods: The modified Hagerman score was applied to 214 male and 86 female patients with mental retardation. The clinical variables in non fragile X and fragile X cases, determined by molecular and cytogenetic methods, were compared. Results: The score in 210 non fragile X males was  $10.5 + 3.7$  (range 3-23), compared to  $21.4 + 2.1$  (range 19 to 23) in the four fragile X patients. All fragile X patients had mental retardation, attention deficits, hyperactivity disorders, hand biting and poor visual contact. Hand biting, flapping and persevering speech were observed in a significantly higher number of fragile X males. Only one of 86 females had fragile X syndrome. Her most releva