Genetic screening to determine an etiologic diagnosis in children with mental retardation Búsqueda de afecciones genéticas como etiología de déficit intelectual en individuos que asisten a escuelas de educación especial

Alliende, M. Angélica

Cámpora, Laura

Curotto, Bianca

Toro, Jessica

Valiente, Alf

Castillo, Marcela

Cortés, Fanny

Trigo, César

Alvarado, Cecilia

Silva, Manuel

Caru, Margarita

Mental retardation or intellectual disability affects 2% of the general population, but in 60% to 70% of cases the real cause of this retardation is not known. An early etiologic diagnosis of intellectual disability can lead to opportunities for improved educational interventions, reinforcing weak areas and providing a genetic counseling to the family. Aim: To search genetic diseases underlying intellectual disabilities of children attending a special education school. Material and methods: A clinical geneticist performed the history and physical examination in one hundred and three students aged between 5 and 24 years (51 males). A blood sample was obtained in 92 of them for a genetic screening that included a standard karyotype, fragile X molecular genetic testing, and search for inborn errors of metabolism by tandem mass spectrometry. Results: This approach yielded an etiological diagnosis in as much as 29 patients. Three percent of them had a fragile X syndrome. Inborn errors of me