

# Williams syndrome: Clinical, cytogenetic, neurophysiological and neuroanatomic features in 44 patients

## Síndrome de Williams: Estudio clínico, citogenético, neurofisiológico y neuroanatómico

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**Background:** Williams syndrome (WS) is a genetically based disorder caused by deletion of elastin and contiguous genes on chromosome 7q11.23. This syndrome is characterized by multiorgan involvement with dysmorphic facial features and a distinctive cognitive profile. It is an interesting model for elucidation of relationships between brain, cognition and genes. Patients have a visual-spatial cognition impaired with relative strengths in social and language abilities. **Aim:** To report clinical, cytogenetic, neurophysiological and neuroanatomic features in 44 patients referred as WS. **Patients and methods:** Forty four patients, aged 2 to 17 years, with the clinical diagnosis of Williams syndrome were studied with fluorescence in situ hybridization (FISH). In three cases, electrophysiological and neuroimaging studies were performed. **Result:** The deletion was confirmed in 23 patients. In three patients with neurophysiological studies, event related potentials suggested a cognitive difficulty i