



Short Communication

Assessment of clinical scoring systems for the diagnosis of Williams-Beuren syndrome

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ABSTRACT. Williams-Beuren syndrome (WBS) is a genetic disorder characterized by physical and intellectual developmental delay, associated with congenital heart disease and facial dysmorphism. WBS is caused by a microdeletion on chromosome 7 (7q11.23), which encompasses the elastin (*ELN*) gene and about 27 other genes. The gold standard for WBS laboratory diagnosis is FISH (fluorescence *in situ* hybridization), which is very costly. As a possible alternative, we investigated the accuracy of three clinical diagnostic scoring systems in 250 patients with WBS diagnosed by FISH. We concluded that all three systems could be used for the clinical diagnosis of WBS, but they all gave a low percentage of false-positive (6.0-9.2%) and false-negative

(0.8-4.0%) results. Therefore, their use should be associated with FISH testing.

Key words: Williams-Beuren syndrome; Clinical diagnosis; Fluorescence *in situ* hybridization; Elastin (*ELN*) gene; Chromosome 7