

Poseidon™ Repeat Free™ Williams-Beuren ELN (7q11) & 7q22 Control probe

Introduction: Williams-Beuren is a congenital syndrome of characteristic elfinlike facies, mental retardation, growth deficiency, cardiovascular anomalies (supraventricular aortic stenosis), and idiopathic infantile hypercalcaemia. The main cause is a contiguous gene deletion of the Williams-Beuren syndrome critical region (WBSCR) that encompasses the Elastin (ELN) gene at 7q11.

Intended use: The Williams-Beuren region probe is optimized to detect copy numbers of the ELN gene region at 7q11. The 7q22 region specific DNA probe at 7q22 is included as control probe.

The probe is recommended to be used in combination with a Poseidon FISH Kit providing necessary reagents to perform FISH (KBI-60002, KBI-60003 or KBI-60001) for optimal results.

Critical region 1 (red): The Williams-Beuren specific DNA probe is direct-labeled with PlatinumBright550.

Control region 2 (green): The 7q22 control DNA probe is direct-labeled with PlatinumBright495.

Reagent: Poseidon probes are direct-labeled DNA probes provided in a ready-to-use format. Apply 10 µl of probe to a sample area of approximately 22 x 22 mm.

Please refer to the Instructions for Use for the entire Poseidon FISH protocol.

Poseidon Repeat Free probes do not contain Cot-1 DNA. Hybridization efficiency is therefore increased and background, due to unspecific binding, is highly reduced.

Interpretation: The Williams-Beuren probe is designed as a dual-color assay to detect deletions at 7q11. Deletions involving the ELN gene region will show one red signal and two green signals at the 7q22 control region (1R2G). Two single color red (R) and green (G) signals will identify the normal chromosome 7 (2R2G).

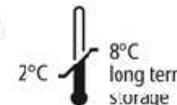
	Normal Signal Pattern	Del 7(q11) ELN
Expected Signals	2R2G	1R2G

References: Mari A et al, 1995, Hum Genet., 96; 444-448
Nickerson et al, 1995, Am.J.Hum.Genet., 56; 1156-1161



Application Manual

KBI-40111
MD Williams-Beuren ELN (7q11) / 7q22



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