ZytoLight® SPEC Williams-Beuren Dual Color Probe

CE IVD

Background

The ZytoLight® SPEC Williams-Beuren Dual Color Probe is designed to detect deletions affecting the chromosomal region 7q11.23 harboring the ELN (elastin, a.k.a. WBS) gene.

The Williams-Beuren syndrome (WBS) is a genetic disorder caused by a hemizygous contiguous gene deletion on chromosome 7q11.23. The estimated prevalence of the disease ranges between 1/7,500 and 1/20,000 newborns.

The WBS deletion region (~1.5-1.8 Mb) consists of a single copy gene region containing app. 28 genes, including the ELN gene that is flanked by repetitive sequences known as low-copy repeats (LCRs).

The deletions arise as a consequence of misalignment of these repetitive sequences during meiosis and a following unequal crossing over due to high similarity of LCRs. Usually, WBS occurs sporadically, but some parents of WBS patients were shown to carry a paracentric inversion of the WBS locus. Presence of this inversion predisposes to chromosomal mispairing in meiosis.

WBS patients clinically display a characteristic pattern of symptoms including vascular stenosis, weakness of connective tissue, a typical face, short stature, overfriendliness, and mental retardation.

FISH analysis can be performed to confirm WBS diagnosis in patients with vascular stenosis together with mental retardation.

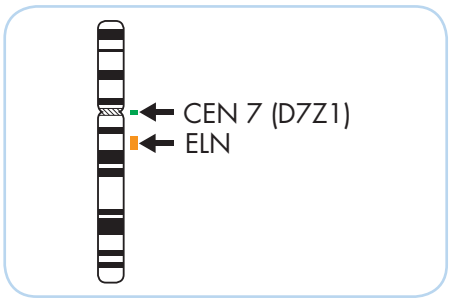
References

Bayés M, et al. (2003) Am J Hum Genet 73: 131-51.
Beuren AJ, et al. (1964) Am J Cardiol 13: 471-83.
Schubert C (2009) Cell Mol Life Sci 66: 1178-97.
Sugayama SM, et al. (2003) Arq Bras Cardiol 81: 462-73.
Williams JC, et al. (1961) Circulation 24: 1311-8.

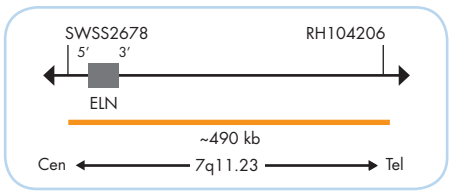
Probe Description

The ZytoLight® SPEC Williams-Beuren Dual Color Probe is composed of:

- ZyOrange (excitation 547 nm/emission 572 nm) labeled polynucleotides (~4.5 ng/µl), which target sequences mapping in 7q11.23** (chr7:73,408,390-73,899,599) harboring the ELN gene region.
- ZyGreen (excitation 503 nm/emission 528 nm) labeled polynucleotides (~4.5 ng/µl), which target sequences mapping in 7p11.1-q11.1 specific for the alpha satellite centromeric region D7Z1 of chromosome 7.
- Formamide based hybridization buffer



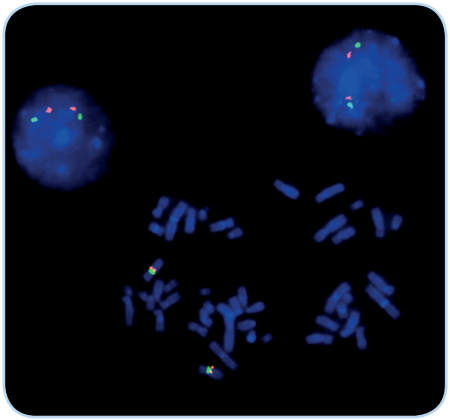
Ideogram of chromosome 7 indicating the hybridization locations.



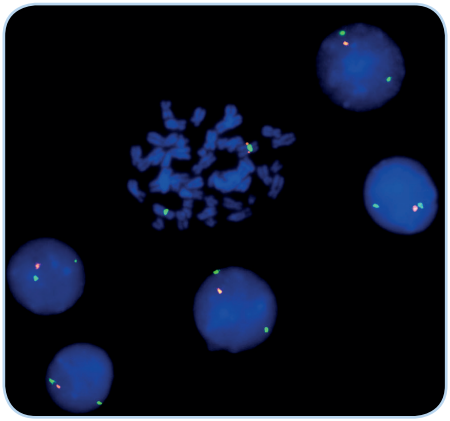
SPEC ELN Probe map (not to scale).

Results

In a normal interphase nucleus, two orange and two green signals are expected. In a cell with deletion of the ELN gene locus, a reduced number of orange signals will be observed.



SPEC Williams-Beuren Dual Color Probe hybridized to normal interphase cells as indicated by two orange and two green signals in each nucleus and to metaphase chromosomes of a normal cell.



Lymphocytes and metaphase chromosomes from a Williams-Beuren syndrome case showing an ELN deletion as indicated by the loss of one orange signal.

Prod. No.	Product	Label	Tests* (Volume)
Z-2302-50	ZytoLight SPEC Williams-Beuren Dual Color Probe CE IVD	●/●	5 (50 µl)
Related Products			
Z-2099-20	ZytoLight FISH-Cytology Implementation Kit CE IVD Incl. Cytology Pepsin Solution, 4 ml; 20x Wash Buffer TBS, 50 ml; 10x $MgCl_2$, 50 ml; 10x PBS, 50 ml; Cytology Stringency Wash Buffer SSC, 500 ml; Cytology Wash Buffer SSC, 500 ml; DAPI/DuraTect-Solution, 0.8 ml		20

** Using 10 µl probe solution per test. IVD labeled products are only available in certain countries. All other countries research use only! Please contact your local dealer for more information.*

***According to Human Genome Assembly GRCh37/hg19*