ZytoLight® SPEC Prader-Willi Dual Color Probe

CE IVD

Background

The ZytoLight® SPEC Prader-Willi Dual Color Probe is designed to detect deletions affecting the chromosomal region 15q11.2 harboring the SNRPN (small nuclear ribonucleoprotein polypeptide N, a.k.a. PWCR) gene.

The Prader-Willi syndrome (PWS) is a sporadic genetic disorder caused by genomic errors that inactivate paternally-inherited genes in the PWS critical region on chromosome 15q11-q13. The absence of expression of one or more of these genes contributes to different phenotypes of PWS. There are three main genetic causes: paternal 5-7 Mb deletion of the 15q11-q13 region, maternal uniparental disomy 15, or imprinting defects in the PWS critical region.

The SNRPN gene is located within the PWS region and has an important regulatory role over the imprinted genes located in chromosome 15.

The estimated prevalence of the disease ranges between 1/15,000 and 1/30,000 newborns. PWS patients clinically display a characteristic pattern of symptoms including hypotonia with poor suck and poor weight gain in infancy, mild mental retardation, hypogonadism, growth hormone insufficiency causing short stature, early childhood-onset hyperphagia and obesity, characteristic appearance, and behavioral and sometimes psychiatric disturbance.

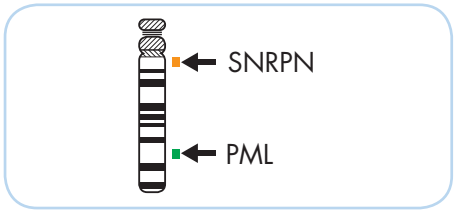
Early diagnosis offers the opportunity to significantly improve health and quality of life of people with PWS. FISH analysis can be performed to detect deletions within the PWS critical region and can help to confirm PWS diagnosis in patients with clinical features characteristic for PWS.

References
Cassidy AB & Driscoll DJ (2009) Eur J Hum Genet 17: 3-13.
Costa RA, et al. (2019) Front Endocrinol (Lausanne) 10: 864.

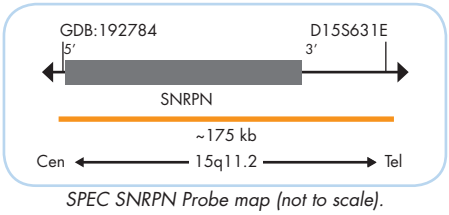
Probe Description

The ZytoLight® SPEC Prader-Willi Dual Color Probe is composed of:

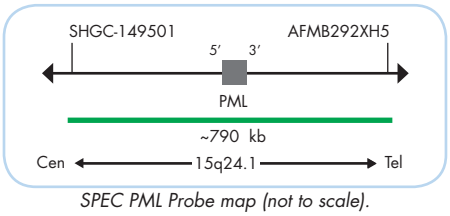
- ZyOrange (excitation 547 nm/emission 572 nm) labeled polynucleotides (~4.5 ng/µl), which target sequences mapping in 15q11.2** (chr15:25,097,811-25,270,969) harboring the SNRPN gene region.
- ZyGreen (excitation 503 nm/emission 528 nm) labeled polynucleotides (~10.0 ng/µl), which target sequences mapping in 15q24.1** (chr15:73,910,690-74,699,298) harboring the PML gene region.
- Formamide based hybridization buffer



Ideogram of chromosome 15 indicating the hybridization locations.



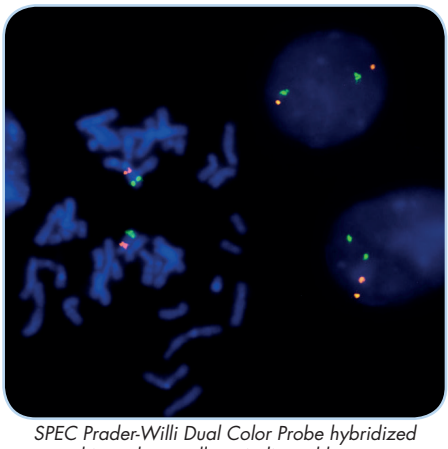
SPEC SNRPN Probe map (not to scale).



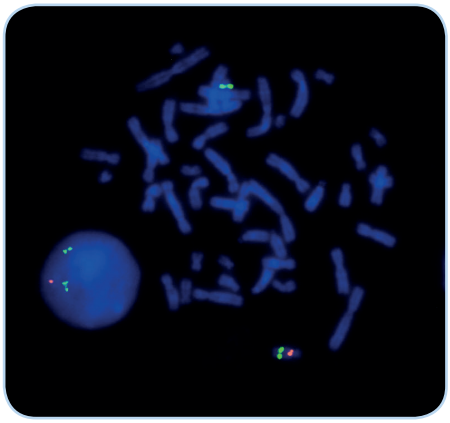
SPEC PML Probe map (not to scale).

Results

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



SPEC Prader-Willi 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 Prader-Willi syndrome case showing deletion affecting the chromosomal region 15q11.2 as indicated by the loss of one orange signal.

Prod. No.	Product	Label	Tests* (Volume)
Z-2318-50	ZytoLight SPEC Prader-Willi 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*