

ATRX/CCPX FISH Probe Kit

Introduction

The ATRX/CCPX FISH Probe Kit is designed to detect the human *ATR*X gene located on chromosome band Xq21.1, along with the number of chromosome X copies per cell. Abnormalities in *ATR*X – also known as *ATR*2, *JMS*, *MRX*52, *MRX*H1, *RAD*54, *RAD*54L, *SFM*1, *SHS*, *XH*2, *XNP* or *ZNF*-H*X* – occur in alpha-thalassemia mental retardation syndrome, which a disorder characterized by severe psychomotor retardation, facial dysmorphism, urogenital abnormalities, and alpha-thalassemia. An essential phenotypic trait is hemoglobin H erythrocyte inclusions.

Intended Use

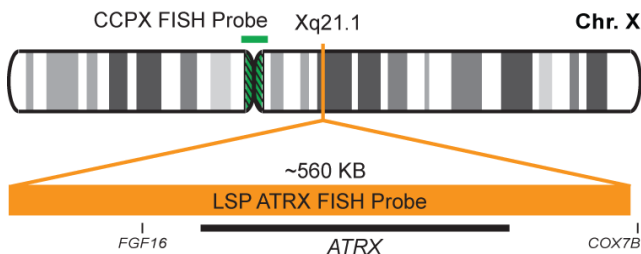
To measure the copy number of the human *ATR*X gene located on chromosome band Xq21.1.

| Cont. | Color |
|-------|-------|
|-------|-------|

LSP ATRX FISH Probe
CCPX FISH Probe

CytoOrange
CytoGreen

Probe Design



LSP ATRX FISH Probe covers a chromosomal region which includes the entire *ATR*X gene. CCPX FISH Probe, derived from chromosome X-specific alpha satellite DNA, is designed to serve as a control to determine the number of chromosome X copies per cell.

Not to Scale

| Cat. No. | Volume |
|----------|--------|
|----------|--------|

CT-PAC118-10-OG

10 Tests (100 µL)

Signal Pattern Interpretation

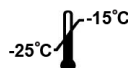
Normal Patterns

2O2G

Abnormal Patterns

Other Patterns

- 1) Sutherland GR, et al. *Am J Med Genet.* 30(1-2):493-508 (1988).
- 2) Gibbons RJ, et al. *Am J Hum Genet.* 51(5):1136-49 (1992).
- 3) Picketts DJ, et al. *Hum Mol Genet.* 5(12):1899-907 (1996).
- 4) Steensma DP, et al. *Blood.* 103(6):2019-26 (2004).
- 5) Nelson ME, et al. *Haematologica.* 90(11):1463-70 (2005).



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* CE IVD only available in certain countries. All other countries are either ASR or RUO. Please contact your local dealer or our headquarters for more information.