BioGenex
Accelerating the pace of precision medicine
48810 Kato Road, Suite 100E & 200E
Fremont, CA 94538
Tel : +1 (800) 421-4149,
Fax: +1 (510) 824-1490,
support@biogenex.com

Data Sheet
eFISH Probes for Chromosome Enumeration

Background

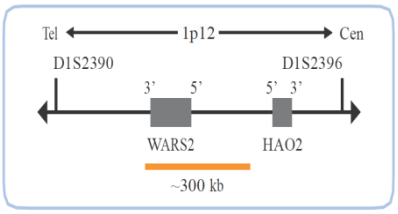
Chromosome Enumeration Probes are designed for identification and enumeration of human chromosomes in interphase cells and as an adjunct to standard karyotyping in metaphases. These probes will produce sharp, bright signals specific for each individual chromosome.

References

Probe Description

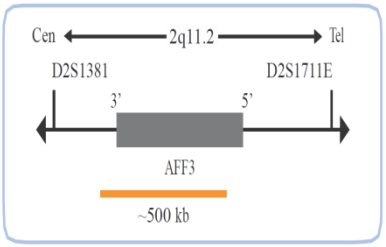
For most chromosomes, direct labeled BioGenex CEN Probes hybridizing to highly repetitive human satellite DNA sequences mainly located at the centromeric regions of chromosomes are applicable. As several chromosomes share the same repetitive sequences resulting in cross-hybridization signals, they cannot be differentiated by centromere specific probes. Instead, these chromosomes can be identified by direct labeled BioGenex eFISH Probes hybridizing in close proximity to the respective satellite DNA sequences or to other chromosome specific loci.

The BioGenex eFISH 1p12 Probe is designed to hybridize in close proximity of centromere 1 at 1p12 harboring WARS2, and HAO2. Since chromosomes 1, 5, and 19 share the same repetitive sequences, they cannot be differentiated by probes detecting centromere specific repeats.



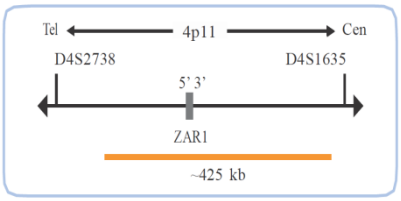
SPEC 1p12 probe map (not to scale)

The BioGenex eFISH 2q11 Probe is specific for the AFF3 (AF4/FMR2 family, member 3) gene region in 2q11.2. Due to cross-hybridizations of chromosome 2 alpha satellites to other centromeric regions, probes specific for 2q11 are frequently used for chromosome 2 copy number detection.



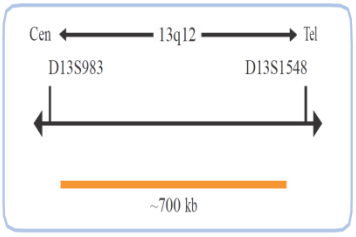
SPEC 2q11 probe map (not to scale)

The BioGenex eFISH 4p11 Probe is designed to hybridize in close proximity of centromere 4 at 4p11 harboring the ZAR1 (zygote arrest 1)gene. For an unambiguous enumeration of chromosome 4 the SPEC 4p11 is found to be more suitable.



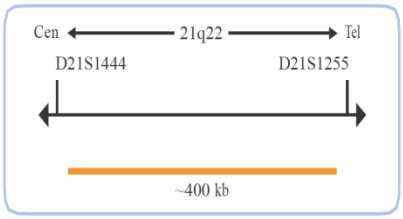
SPEC 4p11 probe map (not to scale)

The BioGenex eFISH13q12 Probe is designed to hybridize in close proximity of centromere 13 at 13q12. Since chromosomes 13 and 21 share the same repetitive sequences, they cannot be differentiated by probes detecting centromere specific repeats.



SPEC 13q12 probe map (not to scale)

The ZytoLight ® SPEC 21q22 Probe hybridizesto the so-called Down Syndrome CriticalRegion on 21q22 commonly duplicated incases with partial trisomy 21.
Since chromosomes 13 and 21 share thesame repetitive sequences, they cannotbe differentiated by probesdetecting centromere specific repeats.



SPEC 21q22 probe map (not to scale)

Result

In a normal interphase nucleus, two signals are expected using Chromosome Enumeration Probes specific for autosomes. Using chromosome Y specific probes will result in normal male cells in one signal and in normal female cells in no signal. Using chromosome X specific probes will

Recommended detection system (Not supplied):		
eFISH Kit	**Cat #**	**Description**
eFISH Histo	DF500-20XE	Automation
eFISH Cyto	DF510-20XE	Automation

BioGenex
Accelerating the pace of precision medicine
48810 Kato Road, Suite 100E & 200E
Fremont, CA 94538
Tel : +1 (800) 421-4149,
Fax: +1 (510) 824-1490,
support@biogenex.com

result in normal male cells in one signal and in normal female cells in two signals per nucleus. Other signal patterns indicate numerical aberrations of the

respective chromosome

Sr#	Enumeration Probes	Cat #	Color
1	eFISH 1p12 Probe	FP084-10X/20X	Orange
2	eFISH 2q11 Probe	FP085-10X/20X	Orange
3	eFISHCEN 3 Probe	FP086-10X/20X	Orange
4	eFISH 4p11 Probe	FP087-10X/20X	Orange
5	eFISHCEN 6 Probe	FP088-10X/20X	Green
6	eFISH CEN 7 Probe	FP089-10X/20X	Orange
7	eFISHCEN 8 Probe	FP090-10X/20X	Green
8	eFISH CEN 9 Probe	FP091-10X/20X	Orange
9	eFISHCEN 10 Probe	FP092-10X/20X	Orange
10	eFISH CEN 11 Probe	FP093-10X/20X	Green
11	eFISHCEN 12 Probe	FP094-10X/20X	Orange
12	eFISH 13q12 Probe	FP095-10X/20X	Orange
14	eFISH CEN 17 Probe	FP097-10X/20X	Orange
15	eFISHCEN 18 Probe	FP098-10X/20X	Orange
16	eFISH 21q22 Probe	FP099-10X/20X	Orange
17	eFISH CEN X Probe	FP100-10X/20X	Orange
18	eFISH CEN Yq12 Probe	FP101-10X/20X	Green

Recommended detection system (Not supplied):		
eFISH Kit	Cat #	Description
eFISH Histo	DF500-20XE	Automation
eFISH Cyto	DF510-20XE	Automation

BioGenex
Accelerating the pace of precision medicine
48810 Kato Road, Suite 100E & 200E
Fremont, CA 94538
Tel : +1 (800) 421-4149,
Fax: +1 (510) 824-1490,
support@biogenex.com

Bibliography

1. Gall, J. G. and Pardue, M. L. (1969). *Proc. Natl. Acad. Sci. USA*63, 378 -383.
2. Rudkin, G. T. and Stollar, B. D. (1977). *Nature* 265,472-473.
3. Hougaard, D. M., Hansen, H. and Larsson, L. I. *(*1997*). Histochem. Cell Biol.* 108*,*335 *-344.*
4. Bauman, J. G., Wiegant, J., Borst, P. and van Duijn, P. *(*1980*). Cell Res.* 128*,*485 *-490.*
5. O'Connor et al. (2008). *Nature Education* 1(1):171.
6. Kievits, T. *et al*. *Cytogenet. Cell Genet.* 53, 134–136 (1990).
7. Antonescu C. (2000).J MolDiagn 2000;2:132-8
8. Germano G, et al.(2010) Cancer Res 70: 2235-44.
9. Kuroda M, et al. (1995). *Am J Pathol.* Nov;147(5):1221–1227

Recommended detection system (Not supplied):		
eFISH Kit	**Cat #**	**Description**
eFISH Histo	DF500-20XE	Automation
eFISH Cyto	DF510-20XE	Automation