

# Data Sheet

## eFISH Probes for Chromosome Enumeration

### Background

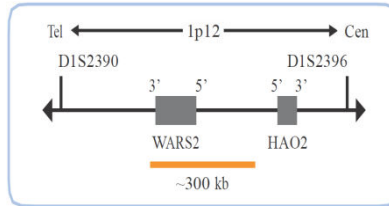
Chromosome Enumeration Probes are designed for identification and enumeration of human chromosomes in interphase cells and as an adjunct to standard karyotyping in metaphases. These probes will produce sharp, bright signals specific for each individual chromosome.

### References

### Probe Description

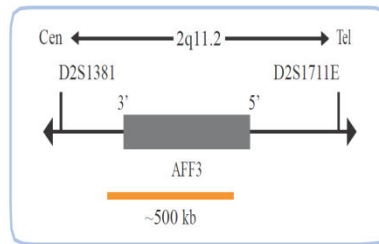
For most chromosomes, direct labeled BioGenex CEN Probes hybridizing to highly repetitive human satellite DNA sequences mainly located at the centromeric regions of chromosomes are applicable. As several chromosomes share the same repetitive sequences resulting in cross-hybridization signals, they cannot 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.



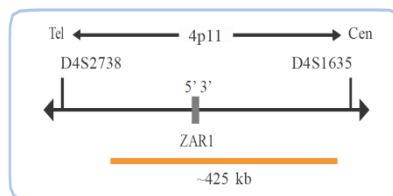
### eFISH 1p12 probe map (not to scale)

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.



### eFISH 2q11 probe map (not to scale)

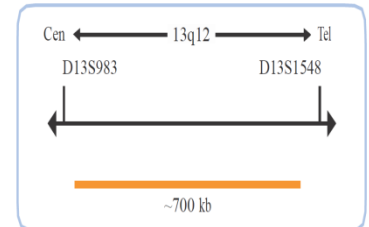
The BioGenex eFISH 4p11 Probe is designed to hybridize in close proximity of centromere 4 at 4p11 harboring the ZAR1 (zygote arrest 1) gene. For an unambiguous enumeration of chromosome 4 the SPEC 4p11 is found to be more suitable.



### eFISH 4p11 probe map (not to scale)

The BioGenex eFISH13q12 Probe is designed to hybridize in close proximity

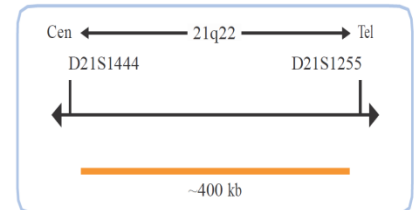
of centromere 13 at 13q12. Since chromosomes 13 and 21 share the same repetitive sequences, they cannot be differentiated by probes detecting centromere specific repeats.



### eFISH 13q12 probe map (not to scale)

The BioGenex® eFISH 21q22 Probe hybridizes to the so-called Down Syndrome Critical Region on 21q22 commonly duplicated incases with partial trisomy 21.

Since chromosomes 13 and 21 share the same repetitive sequences, they cannot be differentiated by probes detecting centromere specific repeats.

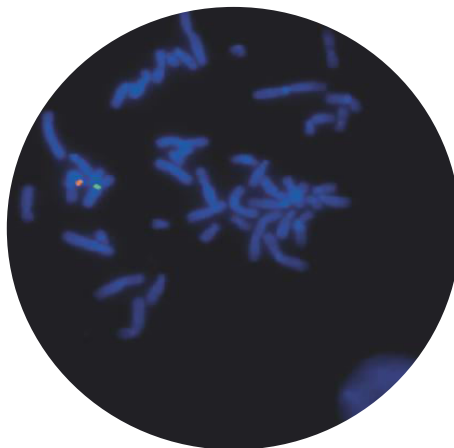


### eFISH 21q22 probe map (not to scale)

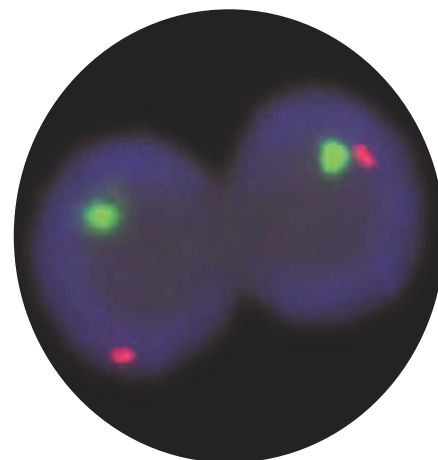
Recommended detection system (Not supplied):		
eFISH Kit	Cat #	Description
eFISH Histo	DF500-20XE	Automation
eFISH Cyto	DF510-20XE	Automation

**Result**

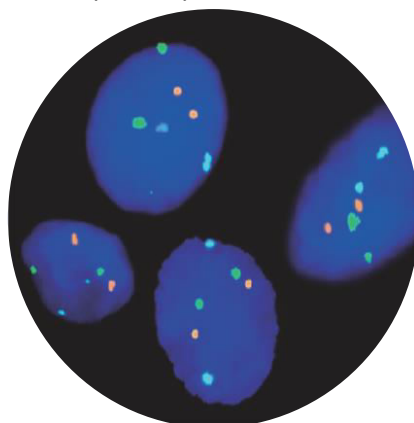
In a normal interphase nucleus, two signals are expected using Chromosome Enumeration Probes specific for autosomes. Using chromosome Y specific probes will result in normal male cells in one signal and in normal female cells in no signal. Using chromosome X specific probes will result in normal male cells in one signal and in normal female cells in two signals per nucleus. Other signal patterns indicate numerical aberrations of the respective chromosome.



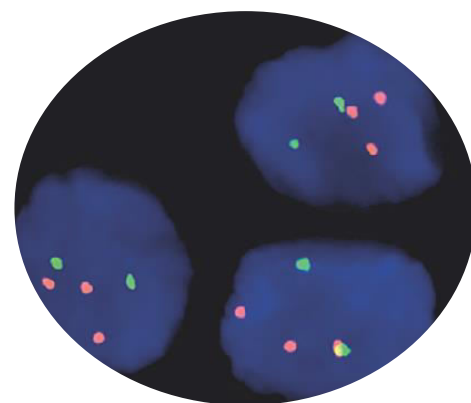
*CEN X/Y Dual Color Probe on a metaphase spread*



*CEN X/Yq12 Dual Color Probe on interphase cells.*



*eFISH 13/CEN 18/21 Triple Color Probe on cytology specimen.*



*eFISH 13/21 Dual Color Probe on interphase cells with trisomy of chromosome 21 (orange).*

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









Sr#	Enumeration Probes	Cat #	Color
1	eFISH 1p12 Probe	FP084-10XE/20XE	Orange
2	eFISH 2q11 Probe	FP085-10XE/20XE	Orange
3	eFISHCEN 3 Probe	FP086-10XE/20XE	Orange
4	eFISH 4p11 Probe	FP087-10XE/20XE	Orange
5	eFISHCEN 6 Probe	FP088-10XE/20XE	Green
6	eFISH CEN 7 Probe	FP089-10XE/20XE	Orange
7	eFISHCEN 8 Probe	FP090-10XE/20XE	Green
8	eFISH CEN 9 Probe	FP091-10XE/20XE	Orange
9	eFISHCEN 10 Probe	FP092-10XE/20XE	Orange
10	eFISH CEN 11 Probe	FP093-10XE/20XE	Green
11	eFISHCEN 12 Probe	FP094-10XE/20XE	Orange
12	eFISH 13q12 Probe	FP095-10XE/20XE	Orange
13	eFISH 13/CEN 18/21 Triple Color Probe	FP096-10XE/20XE	Orange/Green/Blue
14	eFISH CEN 17 Probe	FP097-10XE/20XE	Orange
15	eFISHCEN 18 Probe	FP098-10XE/20XE	Orange
16	eFISH 21q22 Probe	FP099-10XE/20XE	Orange
17	eFISH CEN X Probe	FP100-10XE/20XE	Orange
18	eFISH CEN Yq12 Probe	FP101-10XE/20XE	Green
19	eFISH CEN X/Yq12 Dual Color Probe	FP102-10XE/20XE	Orange/Green

**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

Doc No: 932-FP084-102E; Rev: E; Eff Date: 07-Oct-2021

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

	Temperature Limitation		In Vitro Diagnostic Medical Device
	Use By Date		Batch Code
	Non-Sterile		Consult Instructions for Use
	Representative in the European Community		<b>BioGenex</b>

Doc No: 932-FP084-102E; Rev: E; Eff Date: 07-Oct-2021

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