Table 1. scFISH probes used to detect cytogenetic abnormalities

Chromosome /Disorder	GenBank Accession	Gene	Interval**	Forward PCR Primer Coordinates (Beginning/End)	Reverse PCR Primer Coordinates (Beginning/End)	C+G (%)	CpG (%)	Cytogenetic nomenclature
1/Monosomy 1p36 Sx	AL031282	*CDC2L1*	IVS 11-3′ UTR	9137/9167	13960/13931	65.4	3.9	ish del (1)(p36.3)(CDC2L1-)
	AL031282	*CDC2L1*	3′ UTR	13028/13057	17752/17720	63.4	4.5	
4/ Wolf-Hirschorn Sx	NT_000102	*HD*	Exon 67 – 0.2 kb downstream	267614/267643	271120/271091	55.6	2.4	ish del (4)(p16.3)(HD-)
5/Cri-du-Chat Sx	NT_000149	*CTNND2*	IVS 17	169655/169685	171976/171945	38.6	0.6	ish del (5)(p15.2)(CTNND2-)
	NT_000149	*CTNND2*	IVS 14	199168/199202	203507/203473	42.9	1.1	
	NT_000149	*CTNND2*	IVS 13	212490/212519	216569/216536	40.1	0.3	
	NT_000147	*SEMA5A*	IVS 3	14716/14748	17787/17753	41.6	0.7	ish del (5)(p15.31)(SEMA5A-)
	NT_000147	*SEMA5A*	IVS 3	23905/23935	27710/27676	38.1	0.7	
	NT_000147	*SEMA5A*	IVS 3	30757/30790	33241/33209	41.5	0.8	
	AF119117	*SLC6A3*	IVS 3	28206/28239	31894/31860	62	2.7	ish del (5)(p15.33)(SLC6A3-)[1]
7/Williams Sx	NT_000398	*LIMK1*	IVS 2	31966/31993	35015/34989	62.6	2.6	ish del(7)(q11.23q11.23)(LIMK1-)
	NT_000398	*LIMK1*	IVS 13-3′UTR	59947/59976	62211/62187	61.6	3.3	
8/Langer-Giedeon Sx	NT_002886	*TRPS1*	IVS 1	267731/267760	270758/270724	37.8	1.6	ish del(8)(q23.3q24.1)(TRPS1-)
	NT_002886	*TRPS1*	IVS 1	271242/271271	274437/274404	38.6	0.8	
9/CML (Chronic myelogenous leukemia)	U07561	*ABL1*	Exon 1b-IVS 1b	27182/27213	29388/ 29357	56.3	5.6	ish t(9;22)(q34;q11.2)(ABL st)
	U07562	*ABL1*	IVS 1b	9193/9222	11035/11004	46.8	1.7	
	U07563	*ABL1*	IVS 3	53570/53604	55489/55455	49.8	2.3	ish t(9;22)(q34;q11.2)(ABL mv)
	U07563	*ABL1*	IVS 3	55807/55836	58077/58046	45.9	1.8	
	U07563	*ABL1*	IVS 4-IVS 6	65951/65985	70266/70237	47.5	1.9	ish t(9;22)(q34;q11.2)(ABL mv)
	U07563	*ABL1*	Exon 11-IVS 11	78862/78891	83813/83784	59.5	3.9	
12/ALL (Acute lymphocytic leukemia)	NT_000601	*TEL/ETV6*	IVS2	38216/38245	40091/40062	40.0	1.0	ish t(12;21)(p13;q22)(TEL sp)
	NT_000601	*TEL/ETV6*	IVS3	72543/72564	74385/74361	42.5	1.0	
	NT_000601	*TEL/ETV6*	IVS5-IVS6	95456/95480	97283/97260	43.0	0.8	
13/ Aneuploidy	AL355338	*ZIC2*	~5.8 kb downstream	111114/111145	116046/116012	43.9	1.5	ish del(13)(q32)(ZIC2-) or
	AL355338	*ZIC2*	~2 kb upstream	128595/128627	133039/133006	41.8	1.0	ish del(13)(q32)(ZIC2x3)
15/Prader-Willi, Angelman & Duplication Sx	AC004600	*UBE3A*	IVS 8-IVS 9	41085/41119	45354/45325	35.1	0.6	ish del(15)(q11.2q11.2)(UBE3A-)
	AC004737	*IC/SNRPN*	IVS 3′ to Exon u1B[2]	13740/13769	15414/15387	43.9	3.4	ish del(15)(q11.2q11.2)(IC/ SNRPN-)
	U41384.1	*SNRPN*	Promoter – IVS 1	13906/13930	16116/16086	37.5	0.9	ish dup(15)(q11.2q13)(UBE3A++,
	AC004737	*IC/SNRPN*	IVS 5′-Exon u1B[2]-IVS 3′	31102/31128	33347/33323	38	1.2	IC/SNRPN++), and ish dic(15q11.2q13) (UBE3A++,IC/SNRPN++)
	AC004737	*IC/SNRPN*	IVS 5′-Exon u1B	47792/47821	49470/49441	35.7	0.3	
	AC006596	*MAGEL2*	CDS- 3′ UTR-2 kb downstream	72122/72146	75658/75638	38.9	2.0	ish del(15)(q11.1q11.2)(MAGEL2-) or
	AC006596	*MAGEL2*	~4 kb downstream	76610/76641	78900/78871	39.9	1.2	ish dup (15)(q11.2q11.2)(MAGEL2++)
	AC006596	*MAGEL2*	~22 kb downstream	94501/94535	98601/98567	43.6	1.0	

Chromosome /Disorder	GenBank Accession	Gene	Interval	Forward PCR Primer Coordinates	Reverse PCR Primer Coordinates	C+G (%)	CpG (%)	Cytogenetic nomenclature
16/AML-M4 (Acute Myelogenous leukemia - M4)	NT_000691	*PM5*[3]	~20 kb downstream	24509/24538	27988/27958	66.8	5.2	ish inv(16)(p13q22)(PM5 sp)
	NT_000691	*PM5*[3]	~60 kb downstream	64204/64233	67682/67652	67.1	5.2	
	NT_000691	*PLA2G10*[3] *PKD* *PM5*	IVS 3 IVS 12-Exon 15 ~100 kb upstream	68271/68300	71986/71957	66.9	6.4	ish inv(16)(p13q22)(PLA2G10 mv, PKD mv, PM5 sp)
	NT_000691	*PLA2G10*[3] *PKD* *PM5*	IVS 3 Exon 15-IVS 20 ~100 kb upstream & ~300 kb downstream	71957/71986	75481/75452	66.1	6.0	
	NT_025903	*ABCC1/MDR1*	IVS 6	313783/313812	315675/315645	51.0	2.0	ish inv (16)(p13q22)(ABCC1st)
16/Ruben-stein-Taybi Sx	NT_000671	*CREBBP*	IVS 19 – IVS 20	58653/58685	63854_63823	39.5	1.2	ish del(16)(p13.3)(CREBBP-)
	NT_000671	*CREBBP*	IVS 19	58833/58862	63347/63318	39.2	1.2	
17/Smith-Magenis Sx	NT_000770	*ADORA2B*[4]	Promoter-IVS 1	56443/56472	58524/58491	67.7	9.1	ish del (17)(p11.2p11.2)(ADORA2B-)
	NT_000770	*ADORA2B*[4]	IVS 1	77442/77475	79222/79189	51.8	1.6	
	U80184	*FLI1*	IVS 12-IVS 14	7424/7453	8742/8708	59.1	3.2	ish del (17)(p11.2p11.2)(FLI1-)
	U80184	*FLI1*	IVS 15-Exon 21	9615/9647	11738/11704	60	3.3	
	NT_000760	*MFAP4*	IVS 2 - 3′ UTR	132621/132654	134663/134634	58.3	1.5	ish del(17)(p11.2p11.2)(MFAP4-)
	AL035367	*ZNF179-PAIP1; LLGL/HUGL; SHMT1*	Between ZNF179 PAIP1; IVS7-Exon 13; IVS 4	9818/9850	12272/12241	51.5	1.5	ishdel (17)(p11.2p11.2) (ZNF179/PAIP1/SHMT1-)
	AL035367	*LLGL/HUGL*	Promoter –Exon 1 Promoter – IVS1	1320/1349	5411/5378	57.7	1.9	ish del(17)(p11.2p11.2)(LGLL/HUGL-)
17/Charcot-Marie–Tooth 1A	AC005703	*PMP22*	Promoter (~5 kb upstream)	153173/153202	155027/154994	48.3	1.1	ish dup(17)(p11.2p11.2)(PMP22++)
	AC005703	*PMP22*	IVS 3	176746/176778	179073/179044	46.1	1.0	
	AC005703	*PMP22*[4]	IVS 3	184666/184700	186035/186006	39.3	0.5	
17/ Miller-Dieker Sx	NT_000774	*PAFAH1B1* *EIF-3*[5]	~5kb downstream IVS 24–IVS 27	63645/63679	66603/66573	54.3	3.9	ish del(17)(p13.3)(PAFAH1B1/EIF-3-)
	NT_000774	*PAFAH1B1* *EIF-3*[5]	~7-8 kb downstream IVS 15-IVS 19	68841/68870	71195/71163	49.8	1.0	
	NT_000774	*PAFAH1B1* *EIF-3*[5]	~13 kb downstream IVS 5-IVS 11	75328/75362	78122/78093	43	1.2	
Alagille Sx	AL035456.24	*JAG1*	IVS 2-IVS 3	144875/144904	147028/146995	38.2	0.7	ish del(20)(p12.3p12.3)(JAG1-)
	AL035456.24	*JAG1*	IVS 5-IVS 8	153935/153966	157675/157642	44.2	1.7	
21/Down Sx	AP000160	*DSCR4*	~39 kb upstream	31007/31041	32999/32965	45	0.8	ish (21)(q22.2q22.3)(DSCR4x3)
	AP000160	*DSCR4*	~30 kb upstream	40725/40754	43078/43045	39.2	1.1	
	AP000160	*DSCR4*	~20 kb upstream	49973/50006	52409/52376	37.1	0.5	
21/ ALL	AP000057	*AML1/RUNX*	Promoter-exon 1	98712/98741	102903/102872	47.5	1.4	ish t(12;21)(p13;q22)(AML1 st)
22/ DiGeorge Sx	NT_001039	*HIRA*	IVS 21-IVS 24	819901/819933	823592/823559	53.0	1.4	ish del(22)(q11.2)(HIRA-)
	NT_001039	*HIRA*	IVS 13-IVS 15	843602/843631	846946/846915	52.2	1.1	
	NT_001039	*HIRA*	IVS 12-IVS 13	853946/853975	859116/859085	49.3	1.0	
	NT_001039	*HIRA*	IVS 2-IVS 4	875226/875257	878074/878042	46.7	1.2	

Table 1 (continued)

Chromosome /Disorder	GenBank Accession	Gene	Interval	Forward PCR Primer Coordinates	Reverse PCR Primer Coordinates	C+G (%)	CpG (%)	Cytogenetic nomenclature
22/CML; ALL	U07000	*BCR*	Proximal to major breakpoint in CML	70259/70288	73463/73434	51.6	1.6	ish t(9;22)(q34;q11.2)(BCR st)
	U07000	*BCR*	Proximal to major breakpoint in CML	96305/96336	99705/99675	55.6	2.2	
	U07000	*BCR*	IVS 8	114946/114978	117457/117426	61.4	3.0	ish t(9;22)(q34;q11.2)(BCR mv)
X/ Kallman Sx	NT_001457	*GS2*	Promoter- IVS 2	78970/79000	82994/82960	43.4	2.3	ish del(X)(p22.31)(GS2-)
	AC006062	*KAL1*	IVS 6-IVS 7	38822/38852	42042/42012	38.3	0.8	ish del(X)(p22.31)(KAL1-)
	AC006062	*KAL1*	~8 kb downstream	104433/104465	107097/107072	39.0	0.8	
X/ Turner; Leri-Weill Sx	NT_001151	*SHOX*	IVS 2- exon 3	44615/44646	47505/47473	56.9	8.3	ish del(X)(p22.33)(SHOX-)
	NT_001151	*SHOX*	IVS 4	49637/49669	52251/52217	50.6	3.4	
	NT_001151	*SHOX*	IVS 6	54357/54387	56821/56791	65.3	3.9	
	NT_001159	*TBL1*	IVS 3	175379/175409	179665/179633	42.2	1.7	ish del(X)(p22.22-22.31)(TBL1-)
	NT_001159	*TBL1*	Exon 15- 3′	247264/247293	251290/251257	47.8	2.5	

** WWW hyperlinks are to localized probe sequence in ContigView browser (v9.30a.1; 2 December 2002) at http://www.ensembl.org/Homo_sapiens/

[1] deleted only in Cri-du-Chat patients with terminal deletions, probe is about 2.5 Mb from telomere.
[2] u1B is ~160 kb upstream from the PWS shortest region of overlap and ~85 kb upstream from the AS shortest region of overlap.
[3] PM5 is ~1.3 mb telomeric of MYH11 gene, which is disrupted at the inv(16p) breakpoint. PLA2G10 is ~200 kb telomeric of PM5.
[4] Probe was hybridized in combination with other probes and not individually.
[5] Probe is downstream and adjacent to *PAFAH1B1* gene (formerly known as *LIS1*). An expressed transcript homologous to *EIF-3* is found at these coordinates.