SUPPLEMENTAL

Supplemental1.Total number of CNVs of Unamplified VS Amplified DNA

Sample	Amplified DNA			Unamplified DNA		
	Total CNV	Duplication	Deletion	Total CNV	Duplication	Deletion
1	511	260	251	53	14	39
2	431	189	242	391	156	235
3	170	80	90	301	107	194
4	128	33	95	49	26	23
5	718	407	311	121	69	52
9	42	12	30	23	6	17
10	1008	707	301	27	14	13
11	204	89	115	123	79	44
12	215	31	184	33	18	15
13	235	52	183	301	103	198
16	844	173	671	31	20	11
17	205	29	176	21	1	20

Supplemental 2. Table of patients' information with the corresponding CNVs found in general and affecting miRs

Sample	Gender	Age of Onset	Unamplified DNA Materials					
			total # CNVs	Duplication [total]	Deletion [total]	total # CNVs on miRs	Duplication [miR]	Deletion [miR]
1	F	26	53	14	39	2	2	0
2	F	29	391	156	235	48	17	31 - 1 Complex
3	F	31	301	107	194	24	13	11
4	F	32	49	26	23	7	6	1
5	F	33	121	69	52	12	11	1
6	F	34	29	21	8	3	2	1
7	F	34	100	41	59	11	10	1
8	F	36	60	30	30	6	6	0
9	M	23	23	6	17	2	1	1
10	M	26	27	14	13	8	2	6
11	M	32	123	79	44	11	3	8
12	M	33	33	18	15	5	2	3
13	M	33	301	103	198	25	6	19
14	M	34	9	4	5	2	2	0
15	M	35	89	36	53	7	7	0
16	M	39	31	20	11	5	3	2
17	M	39	21	1	20	2	0	2

Supplemental 3.List of unique deletion of CNV (affecting miR)

Sample	Chr. No	Start Position	End Position	Event	miRNAs	Probe Value	CNV Length	Validation & Method
2	3	49,032,980	49,033,520	CN Loss	hsa-mir-191	-0.30	541	No - q-PCR
2	5	179,374,994	179,375,456	CN Loss	hsa-mir-340	-0.30	463	
2	7	150,566,255	150,566,735	CN Loss	hsa-mir-671	-0.26	481	
2	8	22,158,205	22,158,845	CN Loss	hsa-mir-320a	-0.23	641	No - q-PCR
2	9	130,194,223	130,196,189	CN Loss	hsa-mir-219-2	-0.28	1967	
2	10	134,911,061	134,911,111	Homozygous Copy Loss	hsa-mir-202	-3.45	51	
2	16	30,812,261	30,813,181	CN Loss	hsa-mir-762	-0.40	921	
2	17	72,244,092	72,244,552	CN Loss	hsa-mir-636	-0.48	461	
2	17	76,714,126	76,714,516	CN Loss	hsa-mir-338	-0.34	391	
2	20	32,517,166	32,517,876	CN Loss	hsa-mir-644	-0.27	711	
2	20	61,276,722	61,280,958	CN Loss	hsa-mir-124-3	-2.49	4,236	No - q-PCR
2	22	20,336,838	20,337,698	CN Loss	hsa-mir-301b, hsa-mir-130b	-0.21	861	
2	X	109,185,168	109,188,285	CN Loss	hsa-mir-652	-0.29	3118	No - PCR
3	2	136,139,222	136,139,542	CN Loss	hsa-mir-128-1	-0.28	321	No - q-PCR
3	8	135,881,320	135,883,060	CN Loss	hsa-mir-30b	-0.22	1741	No - PCR/ q-PCR
3	13	52,282,275	52,283,450	CN Loss	hsa-mir-759	-0.29	1176	
11	1	166,611,161	166,611,691	CN Loss	hsa-mir-557	-0.29	531	No - q-PCR
11	14	100,388,375	100,389,552	CN Loss	hsa-mir-770	-0.35	1178	Yes - qPCR
11	14	100,402,602	100,409,513	CN Loss	hsa-mir-493	-0.29	6912	No - PCR
11	14	100,420,202	100,422,248	CN Loss	hsa-mir-432, hsa-mir-136	-0.28	2047	

11	17	53,763,097	53,764,453	CN Loss	hsa-mir-142	-0.28	1357	No - qPCR/PCR
13	1	100,519,380	100,519,780	CN Loss	hsa-mir-553	-0.23	401	
13	9	130,047,066	130,047,226	CN Loss	hsa-mir-3154	-0.53	161	
13	11	110,889,428	110,889,975	CN Loss	hsa-mir-34c	-0.31	548	No - PCR
13	12	52,713,806	52,714,286	CN Loss	hsa-mir-615	-0.29	481	

Supplemental 4.List of recurrent deletion of CNV (affecting miR)

Sample	Chr. No	Start Position	End Position	Event	miRNAs	Probe Value	CNV Length	Validation & Method
10	2	219,574,366	219,574,676	CN Loss	hsa-mir-375	-0.38	310	
2	2	219,574,416	219,574,696	CN Loss	hsa-mir-375	-2.88	280	
10	2	219,574,636	219,574,676	Homozygous Copy Loss	hsa-mir-375	-1.51	41	No -qPCR& PCR
2	2	219,574,636	219,574,696	Homozygous Copy Loss	hsa-mir-375	-2.88	61	No -qPCR& PCR
5	9	72,614,326	72,614,896	CN Loss	hsa-mir-204	-0.30	571	No -qPCR
4	9	72,614,396	72,614,876	CN Loss	hsa-mir-204	-0.30	481	No -qPCR
11	9	72,614,436	72,615,176	CN Loss	hsa-mir-204	-0.48	741	No -qPCR
9	9	72,614,456	72,615,096	CN Loss	hsa-mir-204	-0.22	641	No -qPCR
7	12	96,480,421	96,482,006	CN Loss	hsa-mir-135a-2	-0.24	1586	No -PCR
3	12	96,481,326	96,482,116	CN Loss	hsa-mir-135a-2	-0.30	791	No -PCR
2	14	103,652,470	103,653,860	CN Loss	hsa-mir-203	-0.55	1391	No -qPCR
13	14	103,652,945	103,653,520	CN Loss	hsa-mir-203	-0.46	576	No -qPCR
2	15	39,770,870	39,771,943	CN Loss	hsa-mir-626	-0.28	1074	
13	15	39,771,060	39,771,530	CN Loss	hsa-mir-626	-0.28	471	
3	15	39,771,080	39,771,621	CN Loss	hsa-mir-626	-0.28	542	
2	19	14,501,320	14,501,380	Homozygous Copy Loss	hsa-mir-639	-2.13	61	
13	19	14,501,340	14,501,390	Homozygous Copy Loss	hsa-mir-639	-2.78	51	
13	22	18,400,557	18,400,667	CN Loss	hsa-mir-185	-0.53	111	
2	22	18,400,557	18,400,667	CN Loss	hsa-mir-185	-0.58	111	

Supplemental 5.List of unique duplication CNV (affecting miR)

Sample	Chr. No	Start Position	End Position	Event	miRNAs	Probe Value	CNV Length
2	1	206,041,755	206,042,425	CN Gain	hsa-mir-29c, hsa-mir-29b-2	0.26	671
2	6	126,847,535	126,847,865	CN Gain	hsa-mir-588	0.32	331
2	12	56,500,684	56,504,994	CN Gain	hsa-mir-26a-2	0.26	4311
2	14	100,558,971	100,562,201	CN Gain	hsa-mir-380, hsa-mir-1197, hsa-mir-411, hsa-mir-758, hsa-mir-323, hsa-mir-299	0.23	3231
2	16	55,449,846	55,450,356	CN Gain	hsa-mir-138-2	0.41	511
2	17	25,467,808	25,468,468	CN Gain	hsa-mir-423	0.33	661
2	X	146,088,097	146,088,437	CN Gain	hsa-mir-513b	0.30	341
3	4	8,058,003	8,063,333	CN Gain	hsa-mir-95	0.22	5331
3	14	99,644,821	99,645,900	CN Gain	hsa-mir-342	0.29	1080
3	X	133,507,599	133,508,229	CN Gain	hsa-mir-503	0.25	631
3	X	146,168,452	146,168,962	CN Gain	hsa-mir-514-1	0.26	511
5	3	37,985,954	37,987,500	CN Gain	hsa-mir-26a-1	0.29	1547
5	11	548.089	558.204	CN Gain	hsa-mir-210	0.23	10116
5	19	56,888,322	56,891,079	CN Gain	hsa-mir-125a	0.31	2758
7	16	33,868,749	33,873,931	CN Gain	hsa-mir-1826	0.33	5183
7	20	62,043,153	62,044,223	CN Gain	hsa-mir-1914	0.46	1071
8	17	6,862,043	6,871,965	CN Gain	hsa-mir-497	0.21	9923
8	19	3,912,227	3,912,527	CN Gain	hsa-mir-637	0.52	301

Supplemental 6.List of recurrent duplication CNV (affecting miR)

Sample	Chr. No	Start Position	End Position	Event	miRNAs	Probe Value	CNV Length
3	1	98,284,199	98,284,769	CN Gain	hsa-mir-137	0.26	571
2	1	98,284,209	98,284,840	CN Gain	hsa-mir-137	0.22	632
7	2	132,726,446	132,732,276	CN Gain	hsa-mir-663b	0.39	5831
2	2	132,726,696	132,732,156	CN Gain	hsa-mir-663b	0.27	5461
3	2	132,727,946	132,732,176	CN Gain	hsa-mir-663b	0.29	4231
5	2	132,728,646	132,732,156	CN Gain	hsa-mir-663b	0.96	3,510
16	2	132,728,646	132,741,965	CN Gain	hsa-mir-663b	0.25	13,319
4	2	132,728,696	132,732,206	CN Gain	hsa-mir-663b	1.00	3,510
5	2	219,866,752	219,867,691	CN Gain	hsa-mir-153-1	0.41	940
1	2	219,866,992	219,867,791	CN Gain	hsa-mir-153-1	0.24	800
10	2	219,867,132	219,867,621	CN Gain	hsa-mir-153-1	0.38	490
11	2	219,867,132	219,867,621	CN Gain	hsa-mir-153-1	0.35	490
7	2	219,867,152	219,867,891	CN Gain	hsa-mir-153-1	0.39	740
8	2	219,867,152	219,867,621	CN Gain	hsa-mir-153-1	0.35	470
6	2	219,867,162	219,867,621	CN Gain	hsa-mir-153-1	0.32	460
4	5	93,930,230	93,932,455	CN Gain	hsa-mir-1974	0.51	2226
8	5	93,930,230	93,932,055	CN Gain	hsa-mir-1974	0.39	1826
11	5	93,930,230	93,932,105	CN Gain	hsa-mir-1974	0.31	1876
14	5	93,930,230	93,932,205	CN Gain	hsa-mir-1974	0.43	1976
5	11	1,964,565	1,979,662	CN Gain	hsa-mir-675	0.29	15098
3	11	1,974,410	1,980,312	CN Gain	hsa-mir-675	0.33	5903
12	11	1,974,410	1,975,962	CN Gain	hsa-mir-675	0.26	1553
7	11	1,974,610	1,975,962	CN Gain	hsa-mir-675	0.40	1353

2	12	93,751,430	93,752,720	CN Gain	hsa-mir-492	0.29	1291
13	12	93,752,120	93,752,740	CN Gain	hsa-mir-492	0.26	621
2	13	89,680,812	89,681,902	CN Gain	hsa-mir-622	0.52	1091
13	13	89,680,887	89,681,762	CN Gain	hsa-mir-622	0.40	876
3	13	89,680,952	89,682,157	CN Gain	hsa-mir-622	0.29	1206
7	13	89,681,102	89,682,157	CN Gain	hsa-mir-622	0.24	1056
5	17	7,057,340	7,068,497	CN Gain	hsa-mir-324	0.24	11158
2	17	7,061,615	7,067,755	CN Gain	hsa-mir-324	0.24	6141
3	17	7,066,015	7,073,497	CN Gain	hsa-mir-324	0.25	7483
5	19	51,213,775	51,214,425	CN Gain	hsa-mir-769	0.42	651
2	19	51,213,795	51,214,425	CN Gain	hsa-mir-769	0.35	631
13	19	51,213,795	51,214,065	CN Gain	hsa-mir-769	0.37	271
10	19	51,214,035	51,214,415	CN Gain	hsa-mir-769	0.32	381
7	20	26,136,617	26,137,167	High Copy Gain	hsa-mir-663	0.65	551
7	20	26,136,617	26,138,487	CN Gain	hsa-mir-663	0.79	1,870
4	20	26,136,627	26,138,237	CN Gain	hsa-mir-663	0.38	1611
5	20	26,136,627	26,138,267	CN Gain	hsa-mir-663	0.45	1641
2	20	26,136,727	26,137,117	CN Gain	hsa-mir-663	0.41	391
3	20	26,136,727	26,137,117	CN Gain	hsa-mir-663	0.48	391
12	20	26,136,727	26,137,097	CN Gain	hsa-mir-663	0.49	371
13	20	26,136,727	26,137,117	CN Gain	hsa-mir-663	0.43	391
16	20	26,136,727	26,137,107	High Copy Gain	hsa-mir-663	0.77	381
16	20	26,136,727	26,138,167	CN Gain	hsa-mir-663	0.92	1,440
6	20	26,136,737	26,137,067	CN Gain	hsa-mir-663	0.39	331
3	X	122,523,162	122,523,992	CN Gain	hsa-mir-220a	0.28	831
2	X	122,523,202	122,525,361	CN Gain	hsa-mir-220a	0.35	2160
13	X	122,523,252	122,525,361	CN Gain	hsa-mir-220a	0.24	2110

Supplemental 7.List of recurrent duplication and deletion CNV (affecting miR)

Sample	Chr. No	Start Position	End Position	Event	miRNAs	Probe Value	CNV Length
2	1	153,431,537	153,431,607	Homozygous Copy Loss	hsa-mir-92b	-2.14	71
13	1	153,431,537	153,431,607	Homozygous Copy Loss	hsa-mir-92b	-2.26	71
5	1	153,431,567	153,432,057	CN Gain	hsa-mir-92b	0.42	491
8	1	153,431,617	153,441,587	CN Gain	hsa-mir-92b	0.23	9971
2	2	241,043,266	241,044,246	CN Loss	hsa-mir-149	-2.22	980
15	2	241,044,166	241,044,246	High Copy Gain	hsa-mir-149	2.04	81
2	2	241,044,176	241,044,246	Homozygous Copy Loss	hsa-mir-149	-2.22	71
13	2	241,044,176	241,044,246	Homozygous Copy Loss	hsa-mir-149	-2.29	71
9	3	196,909,527	196,918,180	CN Gain	hsa-mir-570	0.37	8654
3	3	196,909,777	196,911,657	CN Loss	hsa-mir-570	-0.29	1881
7	4	10,979,244	10,979,834	CN Gain	hsa-mir-572	0.29	591
15	4	10,979,454	10,979,584	High Copy Gain	hsa-mir-572	2.05	131
2	4	10,979,464	10,979,584	Homozygous Copy Loss	hsa-mir-572	-1.58	121
4	4	10,979,464	10,979,584	High Copy Gain	hsa-mir-572	0.90	121
13	4	10,979,464	10,979,584	Homozygous Copy Loss	hsa-mir-572	-1.57	121
11	7	30,292,110	30,295,970	CN Gain	hsa-mir-550-1	0.28	3861
3	7	30,294,610	30,296,050	CN Loss	hsa-mir-550-1	-0.26	1441
2	17	1,898,577	1,901,799	CN Loss Complex	hsa-mir-132 & hsa-mir-212	0.21	3222
6	17	1,899,897	1,900,317	CN Loss	hsa-mir-212, hsa-mir-132	-0.61	421
14	17	1,899,897	1,900,357	CN Gain	hsa-mir-212, hsa-mir-132	0.40	461
15	17	1,899,897	1,900,357	High Copy Gain	hsa-mir-212, hsa-mir-132	1.88	461
13	17	1,899,907	1,900,317	Homozygous Copy Loss	hsa-mir-212, hsa-mir-132	-2.03	411
13	17	1,899,907	1,901,049	CN Loss	hsa-mir-212, hsa-mir-132	-0.32	1,142

2	17	1,900,007	1,900,117	Homozygous Copy Loss	hsa-mir-132	-3.21	111
3	17	1,900,017	1,900,107	Homozygous Copy Loss	hsa-mir-132	-1.75	91
7	17	1,900,017	1,910,324	CN Gain	hsa-mir-212, hsa-mir-132	0.28	10308
10	17	1,900,017	1,900,117	Homozygous Copy Loss	hsa-mir-132	-1.67	101
10	17	1,900,017	1,900,367	CN Loss	hsa-mir-132 & hsa-mir-212	-0.62	350
16	17	1,900,017	1,900,117	Homozygous Copy Loss	hsa-mir-132	-1.67	101
12	17	1,900,027	1,900,107	Homozygous Copy Loss	hsa-mir-132	-1.11	81
12	17	1,900,027	1,900,397	CN Loss	hsa-mir-132 & hsa-mir-212	-0.28	370
17	17	1,900,027	1,900,117	Homozygous Copy Loss	hsa-mir-132	-2.49	91
2	17	1,900,207	1,900,317	Homozygous Copy Loss	hsa-mir-212	-2.06	111
5	19	10,689,625	10,691,154	CN Gain	hsa-mir-638	0.25	1530
2	19	10,690,045	10,690,125	Homozygous Copy Loss	hsa-mir-638	-2.69	81
2	19	10,690,045	10,690,315	CN Loss	hsa-mir-638	-0.37	270
4	19	10,690,045	10,690,125	High Copy Gain	hsa-mir-638	0.94	81
10	19	10,690,045	10,690,125	Homozygous Copy Loss	hsa-mir-638	-1.37	81
13	19	10,690,045	10,690,125	Homozygous Copy Loss	hsa-mir-638	-2.65	81
13	19	10,690,045	10,690,285	CN Loss	hsa-mir-638	-0.47	240
15	19	10,690,045	10,690,205	High Copy Gain	hsa-mir-638	2.10	161
16	19	10,690,045	10,690,125	Homozygous Copy Loss	hsa-mir-638	-1.53	81
12	19	10,690,055	10,690,125	Homozygous Copy Loss	hsa-mir-638	-1.07	71
3	19	10,690,065	10,690,125	Homozygous Copy Loss	hsa-mir-638	-1.79	61
3	19	10,690,065	10,690,225	CN Loss	hsa-mir-638	-0.30	160
17	19	10,690,065	10,690,125	Homozygous Copy Loss	hsa-mir-638	-2.86	61
2	22	21,485,270	21,496,090	CN Gain	hsa-mir-650	0.50	10820
11	22	21,495,245	21,495,725	CN Loss	hsa-mir-650	-0.32	481
2	22	21,495,265	21,496,090	High Copy Gain	hsa-mir-650	0.62	826
13	22	21,495,265	21,496,140	CN Gain	hsa-mir-650	0.34	876

3	22	21,495,275	21,496,340	CN Gain	hsa-mir-650	0.32	1066
2	22	36,569,550	36,570,330	CN Loss	hsa-mir-658	-2.60	780
2	22	36,570,250	36,570,330	Homozygous Copy Loss	hsa-mir-658	-2.60	81
10	22	36,570,250	36,570,330	Homozygous Copy Loss	hsa-mir-658	-1.27	81
13	22	36,570,250	36,570,320	Homozygous Copy Loss	hsa-mir-658	-2.53	71
15	22	36,570,250	36,570,330	High Copy Gain	hsa-mir-658	2.02	81
8	X	146,168,452	146,175,963	CN Gain	hsa-mir-514-3, hsa-mir-514-2, hsa-mir-514-1	0.26	7512
4	X	146,168,472	146,177,663	CN Gain	hsa-mir-514-3, hsa-mir-514-2, hsa-mir-514-1	0.27	9192
13	X	146,168,922	146,173,612	CN Loss	hsa-mir-514-2	-0.44	4691
1	X	146,172,752	146,176,313	CN Gain	hsa-mir-514-3	0.21	3562
3	X	146,173,922	146,175,663	CN Gain	hsa-mir-514-3	0.29	1742
13	X	146,173,932	146,176,313	CN Loss	hsa-mir-514-3	-0.44	2382
2	X	152,938,170	152,938,810	CN Loss	hsa-mir-718	-1.27	640
13	X	152,938,210	152,938,810	CN Loss	hsa-mir-718	-1.31	600
3	X	152,938,430	152,938,830	CN Loss	hsa-mir-718	-0.30	401
15	X	152,938,460	152,938,590	High Copy Gain	hsa-mir-718	2.12	131
15	X	152,938,460	152,938,810	CN Gain	hsa-mir-718	1.35	350

CURRICULUM VITAE

Personal Details:

Name: **Avanita S Prabowo**
Current address: Jl. Buring No.30 Malang, East Java, Indonesia 65111
Telephone: +62 8180 3816 999 (mobile)
Email-address: pavanitas@yahoo.com
Date & place of birth: 3 August 1985, Pasuruan, Indonesia
Nationality: Indonesian

Education:

2009-2011 *University of Diponegoro*, Semarang, Indonesia
(Graduated in: Master of Medical Science majoring in Genetic Counseling)

2008-2011 *University of Brawijaya*, Malang, Indonesia
(Graduated *cum laude* in: Master of Health Science majoring in Microbiology)

2003-2007 *The University of Adelaide*, Adelaide, Australia
(Graduated: BSc in Biomedical Science)

1999-2002 *Sacre Coeur*, Glen Iris, Victoria, Australia
(Graduated: Senior High School)

Research Experience:

2010-2011 (6 months) *Biomedical Research Center, University of Brawijaya*
Malang, East Java, Indonesia; Department of Medicine
MSc Internship

Duties included:
- *SDS-PAGE*
- *Protein isolation*
- *Electro-Elution*
- *Bacteria culture*
- *Hemagglutination Assay (HA)*
- *Adhesion test*
- *Enterocytes preparation*
- *Pili cutter*
- *Animal handling (mice)*
- *Polyclonal antibodies production*
- *Western Blot*

2009-2010 (11 months) *Radboud University Nijmegen Medical Centre (RUNMC)*
Nijmegen, The Netherlands; Department of Human Genetics
MSc Internship

Duties included:
- *Micro-RNA microarray*
- *Bioinformatics (Data mining, gene annotation)*
- *PCR*
- *Q-PCR*
- *DNA extraction*

2005-2007 (20 months) *Royal Adelaide Hospital*
Adelaide, SA, Australia; Department of Medicine
BSc & Honours Internship

Duties included:
- *DNA extraction*
- *Microarray*
- *Immunohistochemistry*
- *Histological staining*
- *Apoptosis measurements*
- *Microscopy*
- *Microdissection*
- *Bioinformatics (data mining, pathway generation)*
- *Animal handling (rats)*
- *TUNEL assay*

Awards:

2008-2010 The Excellence Scholarship Program for national and international level
Awarded through the Indonesian Ministry of National Education
http://beasiswaunggulan.depdiknas.go.id/index_en.php

Scientific output:

Publication

-*Prabowo AS*, Suyuti H, Sumarno. Partial characterization of adhesins pili on *Shigella dysenteriae*. Jurnal Kedokteran Brawijaya 2011. (*Accepted*)

-Bowen JM, Gibson RJ, Stringer AM, Chan TW, *Prabowo AS*, Cummins AG, Keefe DMK. Role of p53 in irinotecan-induced intestinal cell death and mucosal damage. *Anti-Cancer Drugs* 2007, 18: 197-210

Abstracts (Oral Presentations)

-Characterization of adhesins pili proteins on Shigella dysenteriae as a potential vaccine candidate.
Avanita Prabowo, Hidayat Suyuti, Wahyudha N Lady, Ali Sabet, Sumarno
The Australian Society for Microbiology, Annual Scientific Meeting, July 4th-8th, 2011 Hobart, Tasmania, Australia.

-Protein pili of 37,8 kDa V.Cholerae Coupled With Cholera Toxin Subunit B V.Cholerae Protect The Intestinal Mice From Efflux Of Water.
Gatoet Ismanoe, Wienarsih, Tri Yudani, *Avanita Prabowo*, Sumarno
The Australian Society for Microbiology Annual Scientific Meeting, July 4th-8th, 2011 Hobart, Tasmania, Australia.

-MicroRNA copy number abnormalities in familial colorectal cancer.
Avanita Prabowo, Ramprasath Venkatachalam, Eveline Kamping, Marjolijn J Ligtenberg, Nicoline Hoogerbrugge, Ad GeurtsVanKessel, Roland P Kuiper.
12th European Workshop on Cytogenetics and Molecular Genetics of Solid Tumours, June 3rd-6th, 2010 Nijmegen, The Netherlands. (Nominated for best presentation).
Published in Cancer Genetics and Cytogenetics Vol.203, 1: 47.

-Gene Expression And Pathway Activation In the Jejunum Following 5-Fu Treatment; Comparison with Irinotecan.
Joanne Bowen, *Avanita Prabowo*, Andrea Stringer, Richard Logan, Rachel Gibson, Dorothy Keefe
International MASCC/ISOO Symposium, june 26th-28th, 2008 Houston, Texas, USA.
Published in Supportive Care in Cancer Vol.16, 6: 619-756

Posters

-Sumbawa fermented horse milk as immunostimulants for 37.8 kDa V.Cholerae vaccine.
Faisal, *Avanita Prabowo*, Kusworini Handono, Wahyudha N Lady, Ali Sabet, Sumarno
The Australian Society for Microbiology, Annual Scientific Meeting, July 4th-8th, 2011 Hobart, Tasmania, Australia.

-Detection of molecule adhesion pili 48 kDa of Salmonella typhi by immunochemistry using sera patients suffering from typhoid fever.
Samsul Islam, Uun Yanuhar, Sanarto Santoso, Sri Winarsih, *Avanita Prabowo*, Sumarno
The Australian Society for Microbiology, Annual Scientific Meeting, July 4th-8th, 2011 Hobart, Tasmania, Australia.

Interest and Abilities:

Interests:	Computers, sports (martial arts, tennis, table tennis, rafting, diving, badminton, softball, soccer), arts, music, travelling and cultures.
Languages:	Indonesian (Native speaker), English (fluent), Javanese (fluent)
Computer applications:	Windows 98/XP/Vista, Office (Access, Excel, Word, Power Point), Open-Office, Photoshop, Vector NTI

PUBLICATION

Abstracts / Cancer Genetics and Cytogenetics 203 (2010) 44–65 47

MICRO-RNA COPY NUMBER ABNORMALITIES IN FAMILIAL COLORECTAL CANCER

Avanita S. Prabowo[1], Ramprasath Venkatachalam[1], Eveline Kamping[1], Marjolijn J. Ligtenberg[1,2], Nicoline Hoogerbrugge[1], Ad Geurts van Kessel[1], Roland P. Kuiper[1]

1. Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen Centre for Molecular Life Sciences, Nijmegen, The Netherlands
2. Department of Pathology, Radboud University Nijmegen Medical Centre, Nijmegen Centre for Molecular Life Sciences, Nijmegen, The Netherlands

The majority of the familial colorectal cancer (CRC) cases cannot be explained by known gene defects, suggesting the existence of other genetic risk factors. In an approach to identify such risk factors, we recently performed a screen for copy number variations (CNVs) in high-risk CRC patients and found, among other lesions, several small deletions affecting microRNA genes. MicroRNAs are negative regulators of approximately 30% of the genes in the human genome, including numerous tumor suppressor genes and oncogenes. In order to comprehensively investigate copy number variation in microRNA genes, and to reveal whether such variations may affect CRC predisposition, we screened for CNVs of microRNA genes in 52 unexplained familial early-onset CRC patients using a custom-made ultimate (tiling) resolution oligo array containing 695 miR genes. We found various small (0.1–5 kb) constitutional deletions and duplications affecting single microRNA genes. Several of these (PCR-validated) CNVs could be detected also in normal controls, whereas others appeared to be rare and were thus far only found in CRC patients. We conclude that CNVs in microRNA genes are more common than previously thought, and that some of them may be associated with CRC predisposition.

NOVEL TRANSLOCATION VARIANT IN EWING SARCOMA INVOLVING THE NFATC2 GENE

Karoly Szuhai[1], Danielle de Jong[1], Hans Tanke[1], Pancras Hogendoorn[2]

1. Department of Molecular Cell Biology, Leiden University Medical Center, The Netherlands
2. Department of Pathology, Leiden University Medical Center, The Netherlands

Ewing sarcoma (ES) is an aggressive sarcoma, and is the second most common bone sarcoma in childhood. Disease-specific t(11;22) (~85–90%), t(21;22) (~5–10%), or rarer variant translocations with the involvement of chromosome 22 (~5%) are present. These translocations result in chimeric genes formed between the EWSR1 gene and a member of the ETS transcription factor family, such as FLI1, ERG. So far, no ES has been identified with a fusion to transcription factors other than ETS. This specificity is important in diagnosing ES. By using a panel of molecular tools such as multicolour FISH and array-CGH, a ring chromosome containing chromosomes 20 and 22 was identified in two index cases with ES-like histological appearance. Molecular karyotyping showed translocation and amplification of regions of chromosomes 20q13 and 22q12. Cloning of the breakpoint showed an in-frame fusion between the EWSR1 and NFATc2 genes. The translocation led to the loss of the N-terminal, calcineurin-dependent control region. Consequently, the remaining intact DNA binding domain of NFATc2 is under control of the EWSR1 promoter region permitting oncogenic activation. Intriguingly, in all cases a distinct histological feature was observed. We conclude that a new translocation involving EWS and NFATc2 has been cloned that is associated with a histological variant of ES. The NFATc2 transcription factor is not a member of the ETS family of transcription factors. NFTAC2 has well-characterized functions in T-cell differentiation and immune response. For the first time a direct involvement of NFATc2 in oncogenesis has been shown. The importance and impact of this finding on molecular diagnosis of small round cell tumors will be discussed.