

Supplementary Table 1. Clinical characteristics of the study population

Sample	Age (years)	GA at amniocentesis (weeks)	Indication of amniocentesis	GA at delivery (weeks)
P1	39	16+3	AMA	35+0
P2	37	17+2	AMA	35+4
P3	33	16+5	Positive screening test for Down syndrome	36+6
P4	35	17+3	Positive screening test for Down syndrome	36+3
P5	32	18+4	Positive screening test for Down syndrome	36+4
N1	31	16+2	Positive screening test for Down syndrome	40+4
N2	35	17+2	Positive screening test for Down syndrome	40+1
N3	39	16+0	AMA	40+5
N4	41	18+2	AMA	40+0
N5	39	17+4	Positive screening test for Down syndrome	40+0

GA, gestational age; AMA, advanced maternal age

In sample names, the prefix P is used for preterm birth and N for normal term birth.

Supplementary Table 2. The candidate gene markers for the preterm birth

Non-normalization						Quantile normalization						RPM normalization						RPKM normalization					
Name	Preterm Birth	Normal Birth	P value (+ FDR)	P value (- FDR)	Fold change	Name	Preterm Birth	Normal Birth	P value (+ FDR)	P value (- FDR)	Fold change	Name	Preterm Birth	Normal Birth	P value (+ FDR)	P value (- FDR)	Fold change	Name	Preterm Birth	Normal Birth	P value (+ FDR)	P value (- FDR)	Fold change
Sorting by p value (+FDR, p < 0.1)																							
MIR6801	2.605	0.021	0.00357	1.47E-07	125.048 up	MIR4420	0.084	1.326	0.00824	2.17E-07	15.836 down	MIR6801	1.399	0.009	0.00215	9.94E-08	163.579 up	MIR6801	62.539	0.024	0.000146	3.84E-09	2582.261 up
LOC105374432	2.759	0.021	0.00357	1.88E-07	132.454 up							SNORA108	0.814	0.009	0.00215	1.13E-07	95.172 up	SNORA108	6.119	0.024	0.000438	2.45E-08	2042.649 up
SNORA108	1.516	0.021	0.00387	3.05E-07	72.754 up							LOC105374432	1.482	0.009	0.00383	3.02E-07	173.267 up	MIR4749	49.434	0.024	0.000438	3.45E-08	2041.176 up
MIR4749	1.574	0.021	0.00471	4.95E-07	94.768 up							MIR4749	1.06	0.009	0.00846	8.89E-07	123.969 up	LOC105374432	6.704	0.024	0.00146	1.54E-07	276.817 up
Sorting by p value (p < 0.05)																							
RDH14	29.913	10.005	0.026	3.41E-06	2.989 up	ARHGAP19	0.153	31.974	0.407	0.000321	209.073 down	ARHGAP19	0.021	17.484	0.68	0.0000893	826.915 down	ARHGAP19	0.045	12.01	0.215	0.0000282	265.690 down
ZNF572	54.524	31.753	0.0784	0.0000145	1.717 up	LOC105375171	29.063	19.499	0.831	0.000127	1.490 up	RDH14	16.062	5.822	0.681	0.000108	2.758 up	RDH14	15.886	5.758	0.68	0.000108	2.759 up
ARHGAP19	0.052	30.044	0.39	0.000148	578.813 down	RDH14	27.736	11.153	0.856	0.000235	2.486 up	LOC102723321	17.767	40.544	0.874	0.000161	2.282 down	LOC102723321	13.923	31.767	0.876	0.000161	2.281 down
GNB4	21.803	10.027	0.39	0.000154	2.174 up	GNB4	19.689	11.808	0.856	0.000263	1.667 up	LOC107987205	2.724	0.02	0.953	0.00041	137.366 up	CHRM5	17.946	10.813	0.961	0.000786	1.659 up
LOC107984025	9.509	2.491	0.505	0.000239	3.816 up	ZNF572	51.559	34.46	0.856	0.000293	1.496 up	IGHMBP2	49.415	28.428	0.953	0.000505	1.738 up	IGHMBP2	17.229	10.172	0.961	0.000902	1.693 up
LOC105375171	31.405	17.36	0.571	0.000318	1.809 up	PTGIR	99.997	68.62	0.938	0.000385	1.457 up	CHRM5	28.698	17.294	0.953	0.000783	1.659 up	RCAN1	42.808	21.374	0.961	0.000934	2.002 up
LOC101927474	19.199	7.039	0.643	0.000703	2.727 up	LOC102723321	30.668	73.252	0.938	0.000561	2.388 down	TMEM225	15.52	6.289	0.953	0.000978	2.467 up	TCF15	4.747	0.058	0.961	0.00103	81.812 up
TMEM225	28.903	10.807	0.643	0.000719	2.674 up	GRN	39.523	19.507	0.938	0.000794	2.026 up	RCAN1	25.769	12.282	0.953	0.00105	2.098 up	BAG3	23.483	15.57	0.961	0.00106	1.508 up
IGHMBP2	92.028	48.848	0.643	0.00079	1.883 up	TMEM225	26.611	12.151	0.938	0.000906	2.189 up	BAG3	40.568	26.901	0.953	0.00106	1.508 up	TMEM225	29.893	11.961	0.961	0.00132	2.499 up
LOC107987205	6.128	0.052	0.643	0.000916	118.056 up	IGHMBP2	88.784	52.238	0.938	0.000996	1.699 up	TCF15	2.848	0.023	0.953	0.00118	125.042 up	LOC107984025	12.244	3.476	0.961	0.00157	3.522 up
LOC105376196	16.027	4.789	0.643	0.000936	3.346 up	SPATA16	11.873	34.209	0.938	0.00121	2.881 down	PTGIR	55.543	37.744	0.953	0.00139	1.471 up	ARPC18	15.108	7.762	0.961	0.0018	1.946 up
TCF15	5.305	0.052	0.643	0.001	102.196 up	MROH5	54.677	32.883	0.938	0.00127	1.662 up	LOC107984025	5.106	1.45	0.953	0.00158	3.521 up	PTGIR	28.864	18.52	0.961	0.00194	1.558 up
LOC101927188	50.404	30.457	0.643	0.0014	1.654 up	TCF15	4.461	0.225	0.938	0.00139	19.786 up	UGT2A3	10.113	22.263	0.953	0.00177	2.201 down	LOC107986124	7.792	15.804	0.961	0.00197	2.028 down
SEMA6B	37.363	12.309	0.643	0.00116	3.035 up	LOC107986385	0.21	4.739	0.938	0.00141	22.540 down	ARPC18	16.907	8.688	0.953	0.0018	1.946 up	LOC107987205	4.463	0.062	0.961	0.00215	17.689 up
MROH5	57.67	29.829	0.643	0.00116	1.933 up	LOC105376196	14.452	5.989	0.938	0.0015	2.413 up	LOC107986124	28.847	58.52	0.953	0.00197	2.028 down	LLGL1	10.155	5.962	0.961	0.0023	1.703 up
RCAN1	47.99	21.104	0.643	0.00119	2.274 up	LOC107986124	50.746	105.374	0.938	0.00157	2.076 down	LLGL1	33.056	19.442	0.953	0.00228	1.700 up	SEMA6B	8.134	2.96	0.961	0.00294	2.748 up
LLGL1	61.562	33.407	0.643	0.0015	1.842 up	UGT2A3	17.084	41.388	0.938	0.00194	2.422 down	AREL1	34.685	21.44	0.953	0.0029	1.617 up	ZNF572	18.412	11.619	0.961	0.00307	1.584 up
GRN	42.373	16.923	0.643	0.00182	2.503 up	LOC105373924	3.632	0.288	0.938	0.00209	12.600 up	ZNF572	29.277	18.479	0.953	0.00308	1.584 up	AREL1	14.816	9.156	0.961	0.0031	1.618 up
AREL1	64.596	36.84	0.643	0.00219	1.753 up	AREL1	61.691	39.648	0.938	0.00215	1.555 up	SEMA6B	20.062	7.163	0.953	0.00309	2.800 up	GNB4	11.443	5.703	0.961	0.00314	2.006 up
LOC105377527	37.025	12.953	0.643	0.00233	2.927 up	SEMA6B	34.721	14.059	0.938	0.00216	2.469 up	GNB4	11.707	5.836	0.953	0.00314	2.006 up	UGT2A3	3.414	7.35	0.961	0.00318	2.153 down
ORA4A7	65.459	33.17	0.643	0.00246	1.973 up	CHRM5	50.497	31.926	0.938	0.00256	1.581 up	SPATA16	7.152	18.795	0.953	0.00366	2.627 down	LOC101927474	6.689	2.658	0.961	0.00384	2.516 up
LOC105376339	17.049	5.346	0.643	0.00262	3.188 up	LOC107987205	4.363	0.225	0.938	0.00274	19.352 up	LOC101927474	10.309	4.096	0.953	0.00384	2.516 up	SPATA16	4.374	12.132	0.961	0.00394	2.773 down
SLCSA2	23.883	7.922	0.643	0.00366	3.014 up	LOC105376339	15.424	6.461	0.938	0.00293	2.387 up	LOC105377527	20.364	7.538	0.953	0.00492	2.701 up	LOC107986385	0.116	5.348	0.961	0.00457	45.959 down
ABCA7	96.441	45.702	0.643	0.00395	2.110 up	RCAN1	44.786	23.062	0.938	0.00303	1.941 up	LOC107986385	0.047	2.626	0.953	0.00529	56.026 down	LOC105377527	10.589	3.919	0.961	0.00491	2.701 up
RNF43	75.138	39.694	0.643	0.00426	1.918 up	LLGL1	58.293	36.014	0.938	0.00318	1.618 up	SLCSA2	12.824	4.61	0.953	0.00661	2.781 up	LOC105373924	5.036	0.124	0.961	0.00646	40.539 up
CHRM5	53.446	29.717	0.643	0.00436	1.798 up	ABCA7	93.675	49.486	0.938	0.00356	1.892 up	LOC105373924	2.286	0.049	0.953	0.00712	47.140 up	SLCSA2	6.252	2.271	0.961	0.00717	2.752 up
BAG3	75.552	46.224	0.643	0.00448	1.634 up	LOC101927188	47.543	33.049	0.938	0.00377	1.438 up	ABCA7	51.784	26.597	0.953	0.00803	1.947 up	ABCA7	9.091	4.675	0.961	0.00732	1.944 up
LOC105373924	4.258	0.098	0.643	0.00465	43.448 up	BRAT1	39.227	19.6	0.938	0.00499	2.001 up	MROH5	30.966	17.36	0.953	0.00865	1.783 up	MROH5	8.019	4.488	0.961	0.00852	1.786 up
LOC107986385	0.098	4.513	0.643	0.00467	46.047 down	ORA4A7	62.324	37.168	0.938	0.00504	1.676 up	LOC105376339	9.154	3.111	0.953	0.00874	2.942 up	LOC105376339	16.059	5.457	0.961	0.00874	2.942 up
LOC105373882	19.877	7.007	0.643	0.00499	2.836 up	LOC107984435	35.84	62.83	0.938	0.00519	1.753 down	LOC102467226	1.273	4.486	0.953	0.00907	3.523 down	LOC102467226	2.526	8.899	0.961	0.00908	3.522 down
BRAT1	41.794	16.972	0.643	0.00517	2.462 up	SLCSA2	21.972	9.627	0.938	0.00539	2.282 up	LOC105376196	8.606	2.787	0.953	0.00985	3.087 up	LOC101927188	11.792	7.722	0.961	0.00983	1.527 up
LOC107986124	53.723	100.555	0.643	0.00519	1.871 down	BAG3	72.11	49.169	0.938	0.00589	1.466 up	LOC101927188	27.065	17.725	0.953	0.00986	1.526 up	LOC105376196	12.711	4.116	0.961	0.00984	3.088 up
PTGIR	103.442	64.856	0.643	0.00522	1.594 up	LOC105373521	0.706	7.099	0.938	0.00589	10.060 down	UGT1A8	5.173	12.711	0.953	0.00994	2.457 down	ARHGAP19-SLIT1	8.984	0.263	0.961	0.0109	34.140 up
UGT2A3	18.834	38.973	0.643	0.00538	2.069 down	LOC107984025	8.44	3.113	0.938	0.00666	2.711 up	LOC105375171	16.863	10.103	0.953	0.0112	1.669 up	LOC105375171	8.805	5.274	0.961	0.0112	1.669 up
LOC102723321	33.088	69.667	0.65	0.00623	2.105 down	LOC101927474	17.451	8.102	0.938	0.00688	2.154 up	GRN	22.753	9.849	0.953	0.0125	2.310 up	UGT1A8	3.348	8.111	0.961	0.0124	2.422 down
SPATA16	13.32	32.295	0.675	0.00739	2.424 down	LOC105377527	35.508	14.065	0.938	0.00714	2.524 up	MIR6778	3.034	0.062	0.953	0.0132	48.604 up	GRN	12.767	5.525	0.961	0.0125	2.310 up
ARPC18	31.487	14.928	0.712	0.00954	2.109 up	UGT1A8	9.349	23.774	0.938	0.00742	2.543 down	LOC107984435	20.506	34.733	0.953	0.0191	1.693 down	MIR6778	141.472	0.544	0.961	0.0196	260.021 up
MIR6778	5.65	0.129	0.712	0.00992	43.693 up	RNF43	72.892	44.093	0.938	0.0102	1.653 up	BRAT1	22.442	9.877	0.953	0.0196	2.272 up	LOC107984435	23.588	45.07	0.961	0.0204	1.910 down
LOC105376787	18.532	6.022	0.712	0.0108	3.077 up	LOC102467226	1.861	8.552	0.938	0.0117	4.595 down	ARHGAP19-SLIT1	17.214	0.256	0.953	0.0203	67.147 up	BRAT1	8.041	3.649	0.961	0.0213	2.203 up
LOC10246722																							

Supplementary Table 3. Validation data of selected differentially expressed genes by qRT-PCR

LOC105374432			RDH14		ZNF572	
	Preterm	Term	Preterm	Term	Preterm	Term
n	21	40	21	40	21	40
Mean	1.777	1.416	1.98	1.36	1.91	1.435
Std. Deviation	1.699	1.193	1.696	1.044	1.819	1.223
Std. Error	0.2622	0.1333	0.2617	0.1167	0.2807	0.1367
95% CI	1.247 to 2.306	1.150 - 1.681	1.451 to 2.508	1.128 to 1.592	1.343 to 2.477	1.163 to 1.707
unpaired two-tailed t-test	0.175		0.014*		0.0889	
one-tailed	0.0873		0.007**		0.0444*	

VOPP1			SERPINA12		TCF15	
	Preterm	Term	Preterm	Term	Preterm	Term
n	21	40	21	40	21	40
Mean	1.941	1.394	1.963	1.391	2.079	1.437
Std. Deviation	1.778	1.157	1.868	1.13	1.801	1.287
Std. Error	0.2743	0.1293	0.2883	0.1264	0.2778	0.1449
95% CI	1.387 to 2.495	1.137 to 1.652	1.380 to 2.545	1.139 to 1.642	1.518 to 2.640	1.148 to 1.725
unpaired two-tailed t-test	0.0425*		0.0373*		0.0252*	
one-tailed	0.0213*		0.0187*		0.0126*	

Supplementary Table 4. Primer sequences for Quantitative Real-Time Polymerase Chain Reaction

Marker name	Gene bank	PCR		
		product size	Forward sequence	Reverse sequence
RDH14	NC_000002.12	70bp	TCTTCTGAACAAATGCCCTCTGA	TCCGTTGAGATTGAGGTAGAAGAGT
SERPINA12	NC_000014.9	70bp	TCCATCTTCAGCTCAGCCTTGT	AGGAGCTTGGCAGACCTTGA
VOPP1	NC_000007.14	70bp	GAGTCTACGGGCCACGATTTAG	CACAGCGACCACCTGTTGTT
ZNF572	NC_000008.11	77bp	ATTCAGCAGCAGCTCTCACCTTA	CGCAGACAGAACATTCATATGGTT
TCF15	NC_000020.11	70bp	AGGTGATCCCATCTCTGATGCT	CCTTCAGGCAGGTAGTTTTTCTG
LOC105374432	NC_000004.12	80bp	CTCGTCTGTTCTCATGCTGCTAA	TGTGAGTCCATTAAACCTCTTTCCT
GAPDH	NM_001256799	292bp	CCTGACCTGCCGTCTAGAAA	GGTGGTCCAGGGGTCTTACT