

Table S1. Gene expression (2- $\Delta\Delta Ct$)

	<i>NIPBL</i>	<i>SMC1A</i>	<i>HDAC8</i>	<i>SMC3</i>	<i>RAD21</i>
Heart	1,00	1,00	1,00	1,00	1,00
Adrenal	3,30	1,45	2,98	1,25	0,65
Salivary	0,89	1,05	0,97	1,34	0,27
Thyroid	6,17	2,02	1,08	2,54	0,83
Kidney	0,14	1,01	1,23	0,89	0,38
Small intestine	5,50	2,81	1,18	2,03	0,53
Stomach	3,26	1,88	1,43	1,78	0,63
Trachea	1,49	1,26	0,97	1,34	0,30
Lung	0,26	1,33	1,19	1,30	0,28
Fetal brain	0,10	1,09	0,91	1,09	0,17
Brain	0,89	1,79	3,25	1,34	3,54
Cerebellum	89,16	8,23	6,56	9,45	3,06
Fetal liver	1,26	9,36	3,25	9,34	1,55
Thymus	41,28	7,83	2,08	6,89	2,36
Spleen	211,25	32,02	21,39	31,20	58,08
Bone marrow	61,18	9,50	1,68	8,05	3,35
Peripheral blood	57,76	7,03	0,89	7,56	2,48

Table S2. Comparison of *NIPBL* expression among original data and public datasets

	Data (2- $\Delta\Delta Ct$)	GTEx (TPM)	FANTOM 5 Project (TPM)	The Human Protein Atlas (TPM)
Heart	1,00	4,833	0,6	2,0
Adrenal	3,30	10,295	-	3,0
Salivary	0,89	15,070	-	2,0
Thyroid	6,17	22,655	-	3,0
Kidney	0,14	5,913	0,6	2,0
Small intestine	5,50	15,410	-	2,0
Stomach	3,26	10,995	-	-
Trachea	1,49	-	-	-
Lung	0,26	20,640	2,0	1,0
Fetal brain	0,10	-	-	-
Brain	0,89	-	1,0	-
Cerebellum	89,16	18,280	3,0	2,0
Fetal liver	1,26	-	-	-
Thymus	41,28	-	-	-
Spleen	211,25	17,455	2,0	-
Bone marrow	61,18	-	1,0	2,0
Peripheral blood	57,76	8,536	-	-

Table S3. Comparison of *SMC1A* expression among original data and public datasets

	Data (2- $\Delta\Delta$ Ct)	GTEx (TPM)	FANTOM 5 Project (TPM)	The Human Protein Atlas (TPM)
Heart	1,00	8,0	19,0	2,0
Adrenal	1,45	13,0	-	3,0
Salivary	1,05	19,0	-	3,0
Thyroid	2,02	23,0	-	3,0
Kidney	1,01	9,0	21,0	3,0
Small intestine	2,81	-	-	3,0
Stomach	1,88	19,0	-	-
Trachea	1,26	-	-	-
Lung	1,33	24,0	12,0	2,0
Fetal brain	1,09	-	-	-
Brain	1,79	-	7,0	-
Cerebellum	8,23	30,0	47,0	2,0
Fetal liver	9,36	-	-	-
Thymus	7,83	-	-	-
Spleen	32,02	27,0	23,0	1,0
Bone marrow	9,50	-	31,0	3,0
Peripheral blood	7,03	7,0	-	-

Table S4. Comparison of *HDAC8* expression among original data and public datasets

	Data (2- $\Delta\Delta$ Ct)	GTEx (TPM)	FANTOM 5 Project (TPM)	The Human Protein Atlas (TPM)
Heart	1,00	9,0	6,0	2,0
Adrenal	2,98	24,0	-	3,0
Salivary	0,97	31,0	-	2,0
Thyroid	1,08	39,0	-	2,0
Kidney	1,23	13,0	10,0	3,0
Small intestine	1,18	37,0	-	3,0
Stomach	1,43	23,0	-	-
Trachea	0,97	-	-	-
Lung	1,19	36,0	7,0	2,0
Fetal brain	0,91	-	-	-
Brain	3,25	-	4,0	-
Cerebellum	6,56	42,0	30,0	1,0
Fetal liver	3,25	-	-	-
Thymus	2,08	-	-	-
Spleen	21,39	39,0	11,0	-
Bone marrow	1,68	-	8,0	1,0
Peripheral blood	0,89	8,0	-	-

Table S5. Comparison of *SMC3* expression among original data and public datasets

	Data (2- $\Delta\Delta$ Ct)	GTEx (TPM)	FANTOM 5 Project (TPM)	The Human Protein Atlas (TPM)
Heart	1,00	2,492	7,0	2,0
Adrenal	2,98	9,960	-	3,0
Salivary	0,97	6,215	-	3,0
Thyroid	1,08	8,913	-	3,0
Kidney	1,23	4,551	14,0	2,0
Small intestine	1,18	4,433	-	2,0
Stomach	1,43	4,553	-	-
Trachea	0,97	-	-	-
Lung	1,19	6,130	11,0	2,0
Fetal brain	0,91	-	-	-
Brain	3,25	-	9,0	-
Cerebellum	6,56	8,354	24,0	1,0
Fetal liver	3,25	-	-	-
Thymus	2,08	-	-	-
Spleen	21,39	5,198	9,0	1,0
Bone marrow	1,68	-	8,0	3,0
Peripheral blood	0,89	1,270	-	-

Table S6. Comparison of *RAD21* expression among original data and public datasets

	Data (2- $\Delta\Delta$ Ct)	GTEx (TPM)	FANTOM 5 Project (TPM)	The Human Protein Atlas (TPM)
Heart	1,00	25,0	-	2,0
Adrenal	2,98	70,0	-	3,0
Salivary	0,97	50,0	-	3,0
Thyroid	1,08	89,0	-	3,0
Kidney	1,23	26,0	-	3,0
Small intestine	1,18	51,0	-	3,0
Stomach	1,43	46,0	-	-
Trachea	0,97	-	-	-
Lung	1,19	78,0	-	3,0
Fetal brain	0,91	-	-	-
Brain	3,25	-	-	-
Cerebellum	6,56	76,0	-	3,0
Fetal liver	3,25	-	-	-
Thymus	2,08	-	-	-
Spleen	21,39	58,0	-	3,0
Bone marrow	1,68	-	-	3,0
Peripheral blood	0,89	28,0	-	-

Table S7. Organs affected in CdLS (frequently in red, intermediate frequency in orange and rarely in yellow) compared to expression levels of causative genes (abundant in red, intermediate in orange and scarce in yellow)

Organ affected	<i>NIPBL</i>	<i>SMC1A</i>	<i>HDAC8</i>	<i>SMC3</i>	<i>RAD21</i>
Brain	0,89	1,79	3,25	1,34	3,54
Cerebellum	89,16	8,23	6,56	9,45	3,06
Stomach	3,26	1,88	1,43	1,78	0,63
Intestine	5,50	2,81	1,18	2,03	0,53
Heart	1,00	1,00	1,00	1,00	1,00
Kidney	0,14	1,01	1,23	0,89	0,38
Liver	1,26	9,36	3,25	9,34	1,55
Lung	0,26	1,33	1,19	1,30	0,28

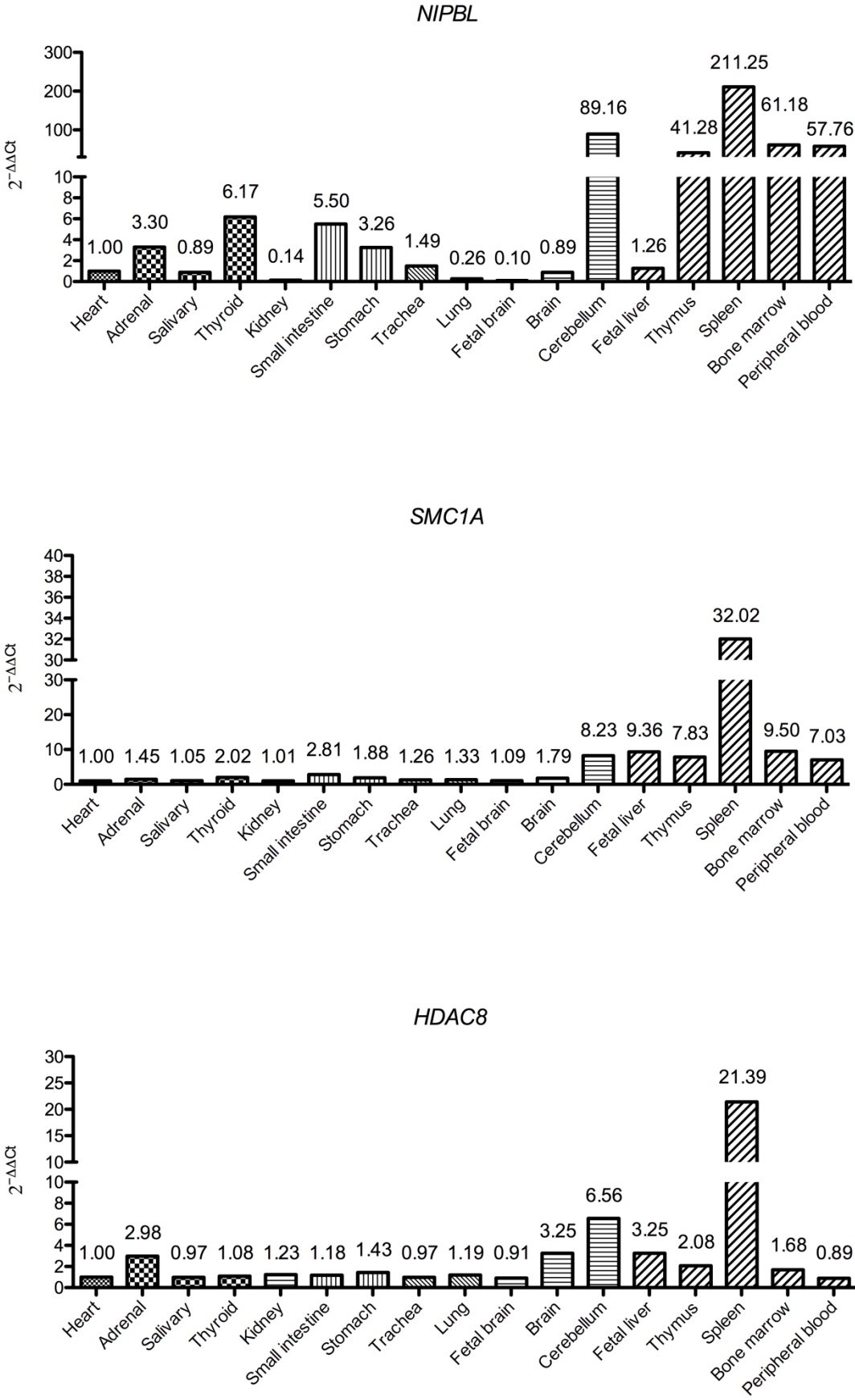
Table S8. Comparison of gene expression in mouse embryos (present data is highlighted in yellow: E13.5 and E15.5) with GXD database (E14.5)

	<i>Nipbl</i>			<i>Smc1a</i>		
	<i>E13.5</i>	<i>E14.5</i>	<i>E15.5</i>	<i>E13.5</i>	<i>E14.5</i>	<i>E15.5</i>
Brain	NO	YES	YES	YES	YES	YES
Cerebellum	YES	YES	YES	YES	YES	YES
Ear	YES	NO	YES	YES	YES	YES
Face	YES	NO	YES	NO	NO	NO
Nose	YES	YES	YES	NO	YES	YES
Tooth	NO	NO	YES	-	YES	YES
Heart	YES	NO	YES	YES	YES	YES
Liver	NO	NO	YES	YES	YES	YES
Lung	YES	NO	YES	NO	YES	YES
Reproductive system	-	YES	YES	-	YES	YES

Table S9. Primers for real-time quantitative-PCR probes

Gene	Forward Primer	Reverse Primer	Probe	Amplicon	Nucleotide Accession Number	Ensembl
<i>NIPBL</i>	ctatgcgaacagcccaaaa	ttcacttgcttactaccattt	55	ctatgcgaacagcccaaaactctctccattcttcttaaa aatgtgttagtaagcaagtgaa	NM_015384.4+NM_133433.3	ENST00000282516.12, ENST00000448238.2
<i>SMC1A</i>	cgacatctagccctgaatctg	attaatgcgagcccaaaagt	78	cgacatctagccctgaatctgcaggaataaaccagctgg agagtgcagtagccaactttgggctctgcattaat	NM_006306.2 + NM_001281463.1	ENST00000375340.10, ENST00000322213.8
<i>SMC3</i>	tgcactgaatgatgagattctg	ttaattcttcatttagcaactgtctg	18	tggagaaaaatccagacaataagagatgctcagcaggatg caagagataaaatggaggatcgaacgccaagt	NM_005445	ENST00000361804.4
<i>RAD21</i>	tcattaagataaagatggcttttcg	tcaaagtcagaaattcttcaggt	38	tcattaagataaagatggcttttcggccaggtgtgttgac ctgcctgaggaaaatcgggaagcagcttataatgccatta ctttacctgaagaatttcagctttga	NM_006265.2	ENST00000297338.6
<i>HDAC8</i>	gcactgcataagcagatgagg	tggagatgctgcagataagc	25	gcactgcataagcagatgaggatagtaagcctaaagtgg cctccatggaggagatggccaccttcacactgatcttat ctgcagcatctcca	NM_001166418	ENST00000439122.6, ENST00000373573.7, ENST00000373589.8, ENST00000373554.5, ENST00000373556.7, ENST00000373559.8, ENST00000373571.5, ENST00000373583.5,

Supplementary Figure S1



Supplementary Figure S2

