

Supplementary Materials for

**Endoluminal biopsy for molecular profiling of human brain
vascular malformations**

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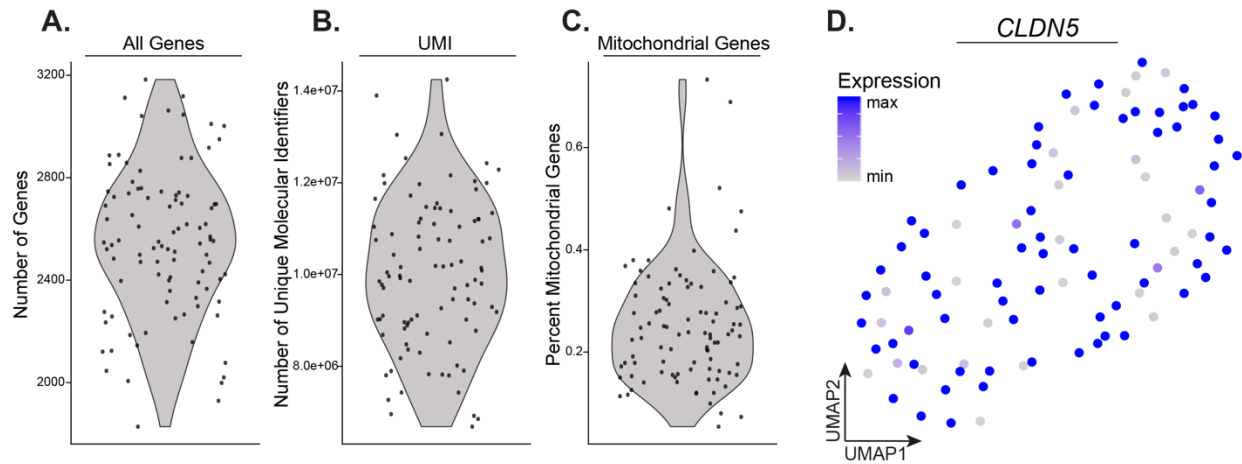
This PDF file includes:

Supplemental Figure 1
Supplemental Table 1-2

Other supplementary material for this manuscript include:

Supplemental Video 1
Supplemental Video 2

Supplemental Figure 1



Supplement Figure 1 – Single cell RNA sequencing of endoluminal biopsied cells following fluorescent-activated cell sorting. (A-C) Quality control violin plots for number of genes (A), number of unique molecular identifiers (B), and percent mitochondrial genes (C) per cell. Each point represents individual cell. UMI, unique molecular identifier. (D) Uniform manifold approximation and projection (UMAP) plot showing distribution of expression of the canonical endothelial marker claudin-5 (encoded by *CLDN5*). Each point represents a single cell. Purple, high expression; Gray, low expression.

Table 1 – Fluidigm quantitative polymerase chain reaction (qPCR) assays.

Gene Symbol	Assay ID
<i>ACTB</i>	GEP00055733
<i>NBPF19</i>	GEP00111261
<i>IFITM1</i>	GEP00110119
<i>FBLX18</i>	GEP00111276
<i>TNFRSF10C</i>	GEP00111262
<i>MARCHF8</i>	GEP00110117
<i>CCND1</i>	GEP00111057
<i>DUSP4</i>	GEP00111271
<i>DUSP6</i>	GEP00111272
<i>EPHA2</i>	GEP00111273
<i>EPHA4</i>	GEP00111274
<i>ETV4</i>	GEP00111064
<i>ETV5</i>	GEP00111270
<i>PHLDA1</i>	GEP00111275
<i>SPRY2</i>	GEP00111268
<i>SPRY4</i>	GEP00111269

Table 2 – Differentially expressed genes in endoluminal sampled endothelium from brain arteriovenous malformations.

Ensembl	Gene Symbol	beta	LRT p-value	FDR-adjusted p-value
ENSG00000155034	FBXL18	4.10645266	4.91E-05	0.049000699
ENSG00000203685	STUM	3.31542487	0.00100392	0.049000699
ENSG00000271383	NBPF19	3.3036345	0.00021954	0.049000699
ENSG00000185885	IFITM1	3.23278956	0.00045122	0.049000699
ENSG00000078804	TP53INP2	2.96350789	0.00077703	0.049000699
ENSG00000073756	PTGS2	2.93603714	0.00094579	0.049000699
ENSG00000173535	TNFRSF10C	2.89827858	0.00012865	0.049000699
ENSG00000163162	RNF149	2.88373808	0.00132133	0.049460607
ENSG00000140718	FTO	2.88055559	0.00053276	0.049000699
ENSG00000165406	MARCHF8	2.87261551	0.00027037	0.049000699
ENSG00000072609	CHFR	2.85322332	0.00055519	0.049000699
ENSG00000111666	CHPT1	2.84516517	0.00026102	0.049000699
ENSG00000184922	FMNL1	2.84276754	0.0012534	0.049460607
ENSG00000173193	PARP14	2.78781901	0.00101537	0.049000699
ENSG00000136279	DBNL	2.76552839	0.00041145	0.049000699
ENSG00000164506	STXBP5	2.66941526	0.00028133	0.049000699
ENSG00000197976	AKAP17A	2.62217897	0.00030916	0.049000699
ENSG00000165312	OTUD1	2.60650648	0.00109386	0.049016269
ENSG00000168763	CNNM3	2.58681786	0.00082695	0.049000699
ENSG00000078699	CBFA2T2	2.52722496	0.00064041	0.049000699
ENSG00000142765	SYTL1	2.50287613	0.0012544	0.049460607
ENSG00000066084	DIP2B	2.47756254	0.00073381	0.049000699
ENSG00000141526	SLC16A3	2.46297948	0.00039547	0.049000699
ENSG00000013725	CD6	2.44259545	0.00061077	0.049000699
ENSG00000181220	ZNF746	2.43561149	0.00041875	0.049000699
ENSG00000175137	SH3BP5L	2.43394932	0.00064138	0.049000699
ENSG00000143772	ITPKB	2.42973179	0.00074902	0.049000699
ENSG00000064547	LPAR2	2.41935966	0.00065458	0.049000699
ENSG00000197555	SIPA1L1	2.41017459	0.00068352	0.049000699
ENSG00000066777	ARFGEF1	2.4074011	0.00052822	0.049000699
ENSG00000135363	LMO2	2.36422975	0.00092616	0.049000699
ENSG00000147138	GPR174	2.35508352	0.00031236	0.049000699
ENSG00000007944	MYLIP	2.35064151	0.00035703	0.049000699
ENSG00000177239	MAN1B1	2.31926212	0.00039947	0.049000699

Ensembl	Gene Symbol	beta	LRT p-value	FDR-adjusted p-value
ENSG00000203879	GDI1	2.31671554	0.00139667	0.049813958
ENSG00000150907	FOXO1	2.30462983	0.00083866	0.049000699
ENSG00000204160	ZDHHC18	2.28868311	0.00077382	0.049000699
ENSG00000176915	ANKLE2	2.28829017	0.00130688	0.049460607
ENSG00000197442	MAP3K5	2.27176119	0.00113885	0.049460607
ENSG00000262156	APOBEC3A	2.2682077	0.00131405	0.049460607
ENSG00000154118	JPH3	2.25924432	0.00029678	0.049000699
ENSG00000173933	RBM4	2.24299596	0.00052092	0.049000699
ENSG00000146859	TMEM140	2.23829547	0.0008354	0.049000699
ENSG00000128011	LRFN1	2.22946825	0.00102955	0.049000699
ENSG00000189077	TMEM120A	2.22242312	0.00086924	0.049000699
ENSG00000152242	C18orf25	2.21042711	0.00103002	0.049000699
ENSG00000120899	PTK2B	2.20375591	0.0008649	0.049000699
ENSG00000100201	DDX17	2.19869226	0.00033081	0.049000699
ENSG00000280404	AC005086.5	2.19384798	0.00131277	0.049460607
ENSG00000058063	ATP11B	2.18630957	0.00103962	0.049000699
ENSG00000188283	ZNF383	2.17744242	0.00063131	0.049000699
ENSG00000125827	TMX4	2.17073543	0.00136164	0.049813958
ENSG00000142409	ZNF787	2.15013386	0.00089606	0.049000699
ENSG00000189339	SLC35E2B	2.14450961	0.00119683	0.049460607
ENSG00000118900	UBN1	2.14264862	0.0011696	0.049460607
ENSG00000101290	CDS2	2.13230713	0.00147506	0.049813958
ENSG00000122068	FYTTD1	2.131432	0.0007025	0.049000699
ENSG00000054967	RELT	2.12781395	0.00099024	0.049000699
ENSG00000110852	CLEC2B	2.12275645	0.00111861	0.049428563
ENSG00000167470	MIDN	2.10742807	0.00149578	0.049882989
ENSG00000204991	SPIRE2	2.10311328	0.00138411	0.049813958
ENSG00000108599	AKAP10	2.10223493	0.00119485	0.049460607
ENSG00000187257	RSBN1L	2.08239597	0.00060265	0.049000699
ENSG00000036549	AC118549.1	2.08239382	0.00056833	0.049000699
ENSG00000166189	HPS6	2.07108829	0.00100479	0.049000699
ENSG00000059378	PARP12	2.0685492	0.00103287	0.049000699
ENSG00000170396	ZNF804A	2.06300355	0.00050989	0.049000699
ENSG00000059758	CDK17	2.05639134	0.0010719	0.049016269
ENSG00000142197	DOP1B	2.05243989	0.00123015	0.049460607
ENSG00000067221	STOML1	2.04454439	0.00030313	0.049000699
ENSG00000143226	FCGR2A	2.03221794	0.0005067	0.049000699
ENSG00000090924	PLEKHG2	2.01599822	0.00093373	0.049000699

Ensembl	Gene Symbol	beta	LRT p-value	FDR-adjusted p-value
ENSG00000153048	CARHSP1	1.987659	0.00062668	0.049000699
ENSG00000136937	NCBP1	1.96564919	0.00137847	0.049813958
ENSG00000161980	POLR3K	1.94395534	0.00132921	0.049460607
ENSG00000179111	HES7	1.93821954	0.000666	0.049000699
ENSG00000162086	ZNF75A	1.92170893	0.00053583	0.049000699
ENSG00000169429	CXCL8	1.92085674	0.00102084	0.049000699
ENSG00000126804	ZBTB1	1.9164277	0.00117222	0.049460607
ENSG00000127528	KLF2	1.90517613	0.00028713	0.049000699
ENSG00000136874	STX17	1.89637098	0.00086504	0.049000699
ENSG00000166337	TAF10	1.88467234	0.00145053	0.049813958
ENSG00000140941	MAP1LC3B	1.8693981	0.00040446	0.049000699
ENSG00000163884	KLF15	1.85490818	0.00063312	0.049000699
ENSG00000120129	DUSP1	1.79797747	0.00147845	0.049813958
ENSG00000131873	CHSY1	1.79505295	0.00103869	0.049000699
ENSG00000117425	PTCH2	1.7719279	0.00015292	0.049000699
ENSG00000241258	CRCP	1.75382857	0.00128127	0.049460607
ENSG00000172000	ZNF556	1.745828	0.00067889	0.049000699
ENSG00000080947	CROCCP3	1.72520817	0.00062101	0.049000699
ENSG00000162104	ADCY9	1.72071956	0.00141877	0.049813958
ENSG00000230254	HLA-E	1.69757303	0.00041121	0.049000699
ENSG00000201822	RNA5SP149	1.67883657	0.00054477	0.049000699
ENSG00000237022	HLA-C	1.67857097	0.00059431	0.049000699
ENSG00000066336	SPI1	1.67827955	0.00078246	0.049000699
ENSG00000160877	NACC1	1.63509686	0.00108068	0.049016269
ENSG00000105392	CRX	1.62112921	0.00144805	0.049813958
ENSG00000133316	WDR74	1.57507282	0.00025744	0.049000699
ENSG00000203668	CHML	1.53559601	0.00109541	0.049016269
ENSG00000249709	ZNF564	1.50743077	0.00147962	0.049813958
ENSG00000233388	AL022334.1	-1.0924709	0.0012334	0.049460607
ENSG00000119335	SET	-1.1846343	0.00095183	0.049000699
ENSG00000101972	STAG2	-1.3926799	0.00092366	0.049000699
ENSG00000155008	APOOL	-1.528255	0.00037111	0.049000699
ENSG00000171453	POLR1C	-2.105712	0.00049755	0.049000699
ENSG00000162989	KCNJ3	-3.2632673	0.00075131	0.049000699

LRT, log-ratio test; FDR, false-discovery rate; beta, approximation of log-fold change