

Supplementary material

Supplementary Table. Diagnostic techniques performed in the autopsy and biopsy cohorts from the Pitié-Salpêtrière and Bicêtre Hospitals.

Cases			MRI <i>ante-mortem</i>	MRI <i>post-mortem</i>	Image findings	RT-PCR	ISH	RNAscope ISH	NGS	EM	IHC	Immunofluorescence					
											SP	SP	BIP	Giantin	Furin	TGN46	Cath B
PSL 1	biopsy	patient # 1	yes	no	Microbleeds	yes	no	no	yes	no	yes	no	no	no	no	no	no
PSL 2	biopsy	patient # 2	yes	no	Microbleeds	yes	no	no	yes	no	yes	no	no	no	no	no	no
PSL 3	biopsy	patient # 3	yes	no	Microbleeds	yes	no	no	yes	no	yes	no	no	no	no	no	no
PSL 4	autopsy	patient # 4	yes	yes	Microbleeds	yes	yes	yes	yes	yes	yes	yes	yes	yes	yes	yes	yes
PSL 5	autopsy		yes	yes	Deep venous thrombosis	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 6	autopsy		no	yes	Deep venous thrombosis	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 7	autopsy		no	yes	Deep venous thrombosis	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 8	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 9	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 10	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 11	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 12	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
PSL 13	autopsy		no	yes	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
BH 14	autopsy	patient # 5	yes	no	Microbleeds	no	no	no	no	no	yes	yes	no	yes	yes	yes	yes
BH 15	autopsy	patient # 6	no	no	Microbleeds	no	no	no	no	no	yes	yes	no	yes	yes	yes	yes
BH 16	autopsy		no	no	No CNS involvement	no	no	no	no	no	yes	yes	no	yes	yes	yes	yes
BH 17	autopsy		no	no	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
BH 18	autopsy		no	no	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
BH 19	autopsy		no	no	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no
BH 20	autopsy		no	no	No CNS involvement	no	no	no	no	no	yes	no	no	no	no	no	no

BH: Bicêtre Hospital; **EM:** Electron microscopy; **IHC:** immunohistochemistry; **ISH:** *In-situ* hybridization; **MRI:** Magnetic Resonance Imaging; **NGS:** Next Generation Sequencing; **PSL:** Pitié-Salpêtrière Hospital; **RT-PCR:** Real-time polymerase chain reaction.



Supplementary Figure

