Supplementary Table 3 **DupAPP clinical and pathological summary**[1-11]

Reference	Affected individuals	Size of duplication	Age of onset of AD/ ICH (years)	Seizures	ICH/ strokes	Neuropathology
Rovelet-Lecrux et al, 2006	5 kindreds (14 confirmed cases) France	0.58 Mb – 6.37 Mb	42 – 59 (in all confirmed cases)	Not reported	4 / 14 (29%)	5 cases from 3 kindreds AD + severe CAA; micro infarcts in 3
Sleegers et al, 2006	1 kindred (4 confirmed cases) and 1 other proband Netherlands	0.7 Mb	53 – 62 (in all confirmed cases)	3 / 4 (75%)	several in kindred had strokes	1 case – AD + CAA
Rovelet-Lecrux et al, 2006 & Rovelet-Lecrux et al 2007	1 kindred, 14 affected individuals Finland	0.55 Mb	40 – 54 (in all affected cases)	2 / 14 (14%)	5 / 14 (36%)	3 cases - AD + CAA
Thonberg et al, 2011	1 proband Sweden	1.09 Mb	55	Not reported	-	-
Kasuga et al, 2011	2 probands Japan		52 – 53	None	1 / 2 (50%, on CT scan)	-
Wallon et al, 2012	7 kindreds, 19 affected individuals France	0.83 Mb – 14.7 Mb	41 – 64 (in all affected cases)	16 / 19 cases (84%; index symptom in 2)	Cases in all 7 kindreds	5 cases - moderate to severe AD + severe CAA 1 case cortical Lewy Bodies
McNaughton et al, 2012	5 probands UK	1.6 – 6.6 Mb; one with interrupted duplication of 15.5 Mb	39 – 61 (in all affected probands)	4/4 (100%)	1 / 5 (20%)	-
Hooli et al, 2012 ; Hooli et al, 2014	2 kindreds, 5 affected individuals USA	0.38 Mb (APP only) & 3.4 Mb	43 – 52 years; one case with no disease at age 60	-	-	-
Swaminathan et al, 2012	1 proband USA	0.5 Mb	59	Not reported	Not reported	Not reported
Llado et al, 2014	1 proband Spain	14.5 Mb	54	-	Yes (presented with ICH)	AD + severe CAA

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