

Figure S3: Positive Predictive Value calculation

The PPV value was calculated using a MedCalc based interface (https://www.medcalc.org/calc/diagnostic_test.php)

Diagnoses were based on the DNA methylation profile of the two markers:

- Marker 1: scaffold33832:13205..13768, considered as D for diagnosis when the threshold is above 25% (otherwise SWC)
- Marker 2: scaffold1255:365901..366597, considered as D for diagnosis when the threshold is equal to 0% (otherwise SWC)

True positive took into account a positive occurrence of D diagnosis in the two markers in the D group otherwise they were considered as false negative.

False negative took into account a positive occurrence of D diagnosis in the two markers in the control group otherwise they were considered as true negative.

Results from the SWC group:

Individuals from SWC group	1	2	3	4	5	6	7	8	9	10
% methylation in marker 1 (threshold = 25%)	0,00	29,17	62,50	0,00	0,00	0,00	20,83	0,00	45,83	20,83
Diagnosis in marker 1	SWC	D	D	SWC	SWC	SWC	SWC	SWC	D	SWC
% methylation in marker 2 (threshold = 0%)	29,76	29,83	0,00	0,00	0,00	14,43	28,58	30,72	28,43	0,00
Diagnosis in marker 2	SWC	SWC	D	D	D	SWC	SWC	SWC	SWC	D
Final diagnosis (marker 1 + marker 2)	SWC	SWC	D	SWC	SWC	SWC	SWC	SWC	SWC	SWC

Results from the D group:

Individuals from D group	21	22	23	24	25	26	27	28	29	30
% methylation in marker 1 (threshold = 25%)	41,67	41,67	16,67	41,67	54,17	12,50	20,83	50,00	12,50	50,00
Diagnosis in marker 1	D	D	SWC	D	D	SWC	SWC	D	SWC	D
% methylation in marker 2 (threshold = 0%)	0,00	0,00	0,00	0,00	0,00	0,00	0,00	0,00	0,00	0,00
Diagnosis in marker 2	D	D	D	D	D	D	D	D	D	D
Final diagnosis (marker 1 + marker 2)	D	D	SWC	D	D	SWC	SWC	D	SWC	D

	Present	n	Absent	n	Total
Positive	True positive	a	False positive	c	a+c
Negative	False Negative	b	True negative	d	b+d
Total		a+b		c+d	
PPV	a/(a+c)				
NPV	d/(b+d)				

	Present	n	Absent	n	Total
Positive	True positive	6	False positive	1	a+c
Negative	False Negative	4	True negative	9	b+d
Total		a+b		c+d	
PPV	85.71				
NPV	69.23				