

1 **Supporting information**

2 **S1 Table:** Quality analysis comparing the sample with block-IV and-V questions completed

3 (n=524) to that with these questions uncompleted (n=281)

		Sample 281 n (%)	Sample 524 n (%)	p value (Chi- squared test)
Delay	Diagnosis within 1 year	100 (35.6)	181 (34.5)	0.767
	Diagnostic delay	181 (64.4)	343 (65.4)	
Sex	Men	105 (37.3)	201 (38.3)	0.782
	Women	176 (62.6)	323 (61.6)	
Type of RD	Osteomuscular	20 (7.1)	60 (11.4)	0.138
	Nervous system	92 (32.7)	155 (29.5)	
	Congenital malformations	40 (14.2)	61 (11.6)	
	Ocular	59 (20.9)	95 (18.1)	
	Endocrine	17 (6)	49 (9.3)	
	Other	53 (18.8)	104 (19.8)	
Age at diagnosis		n (\bar{x})	n (\bar{x})	p value (Mann- Whitney U Test)
		277 (38.5)	524 (37.1)	0.053

4