

Supplementary Table 3. Phenotypic data for hEDS Patients.

Patient ID	Age	Sex	Beighton score	cEDS	vEDS	hEDS	kEDS	Other features	Vascular/ cardiac complications	GI Symp	Dys-Autonomia	Family History
				Major criteria Minor criteria	Major criteria Minor criteria	Major criteria Minor criteria	Major criteria Minor criteria					
61	30-39	F	–	C	–	H	–	–	–	–	–	Son: hEDS Sister: hEDS, COL3A1:VUS
65	60-69	F	3	C	E	H	–	–	Aneurysm, NOS	–	+	Mother: ICA Paternal grandmother: Cerebral Hemorrhage Paternal uncle: Cerebral Hemorrhage
70	10-19	M	4	C	–	H	–	ejection systolic click	–	–	–	Mother: hEDS Maternal grandmother: OA, GH; Umbilical hernia
74	50-59	F	–	C	–	H	–	Str	–	–	–	Brother: PXE Mother: AA-NOS, Bru, VV
100	50-59	F	7	A, C	E	H, I	–	–	ICA	–	–	Brother: GH Daughter: GH
107	40-49	M	4	–	E	H, I	–	–	–	–	–	Paternal grandfather: AA-NOS Sister: hEDS Paternal cousin 1: TAD. Paternal cousin 2: AoR.
191	30-39	F	3	C	–	H, I	–	MVP	–	–	–	Mother: GH Daughter: GH Son: GH, GORD
374	50-59	M	–	C	–	H	–	MVP aortic valve surgery	–	–	–	N/A
385	30-39	F	–	C	–	H, I	–	MVP	–	–	+	Father: ICA Paternal grandmother: ICA, AAA
395	50-59	M	–	A, C	–	H, I	–	–	–	–	–	Daughter: MVP, GH, SS, HS, BS
397	20-29	F	–	A, C	–	H, I	–	MVP	–	+	–	Mother: cEDS/ hEDS overlap Father: hEDS
402	30-39	M	6	A, C	–	H, I	–	–	–	–	–	Father: GH, TS Sister: Knee dislocation, GH, Heart murmur
404	40-49	M	9	A, C	–	H, I	–	–	–	–	–	Mother: GH Father: GH Paternal grandmother: AA-NOS Paternal grandfather: AA-NOS Daughter: hEDS
428	60-69	F	–	B, C	D, F	H, I	–	Poa	–	–	–	Daughter: hEDS
475	30-39	F	7	–	–	H, I	–	–	–	–	+	Daughter: PP, GH Son 1: GH, IF Son 2: GH, Ftg
495	40-49	F	6	C	–	H	–	PGMW OP Bradycardia	–	–	–	Daughter: PGMW, UI Mother: PGMW, UI Sister 1: PGMW Sister 2: PGMW, VV Sister's 2 children: GH Maternal aunt: PGMW, UI, VV
536	40-49	M	1	A, B	D	I	–	Dilated cardiomyopath	–	–	–	–
560	20-29	F	5	A, C	–	H, I	–	–	–	+	–	Mother: hEDS Sister: hEDS, Filamin A gene mutation in exon 48 (de novo) Maternal Grandmother: GH
566	60-69	M	4	A, C	E	H, I	J	OP VV	–	–	–	Father: TS, My Mother: My
584	20-29	F	–	A, C	–	H, I	–	PE	–	–	–	Son 1: hEDS, TS, pneumothorax Son 2: GH, Hyperextensible skin, PE
612	30-39	F	7	C	–	H	–	–	–	–	–	Daughter: hEDS
621	20-29	F	6	A, B	–	H, I	–	Palpitations	–	–	–	Mother: GH, Maternal aunt: GH, Sister (identical twin): GH
630	30-39	F	7	C	–	H, I	–	PGMW MVR, Aortic regurgitation; Tricuspid regurgitation	–	+	–	Father: GH, TS Paternal grandfather: GH, TS Paternal great grandfather: GH, TS
638	40-49	F	–	C	–	H, I	–	tall stature	–	–	+	Sister: hEDS Father: TS
650	30-39	F	7	C	–	H	–	Livedo reticularis	–	+	–	Fhx of GH Maternal aunt: Pulmonary artery atresia

669	20-29	F	7	A, C d, g, i	E	H, I s, u	-	PMGW	-	-	-	Mother: PGMW Sister: GJH, PGMW Daughter: hEDS
670	30-39	F	8	B, C d, e, f, g, h, i	D	H, I u	-	PMGW	-	-	-	Mother: PGMW, GJH Father: SS, Dupuytren's contracture Daughter: Goldenhaar syndrome, GJH Son: GJH, Cleft palate
673	50-59	M	3	C g	D	H u	-	-	AoR	-	-	Son: GJH Sister: GJH
681	50-59	F	-	A, C -	-	H, I t, u	J, L v, y	TS PGMW	-	+	-	Mother: GJH Father: Aortic aneurysm
682	40-49	F	6	A, C g, i	E q	H, I t, u	-	Pd	-	+	-	Mother: GJH, Pd Father: GJH, Pd Brother: GJH, Pd Sister: GJH, Pd Maternal aunt: GJH, Pd
703	10-19	F	-	C -	-	H t, u	-	-	-	-	-	-
755	40-49	F	4	A, C d, e	-	H, I u	J, K	-	-	+	-	Father: TS 2 daughters: GJH, CHD
761	20-29	M	6	B, C d, f	-	H, I t, u	J v	tall stature tibial bowing Sco	-	-	+	Mother: GJH, Maternal cousins: GJH
769	20-29	F	3	C d, g	-	H s, t, u	-	brachydactyly	-	+	-	Mother: GJH Maternal mother: GJH Maternal grandmother: GJH Maternal great grandmother: GJH Maternal aunt: GJH Father: GJH, brachydactyly Paternal grandmother: OA, OP
778	20-29	F	7	A, C d, i	-	H, I s, t, u	-	Palpitations	-	+	+	Mother: GJH, Cerebral Hemorrhage Maternal grandmother: GJH Children: GJH
781	40-49	F	5	A, C f	E	H, I t, u	-	-	ICA	+	-	Father: GJH, Hyperextensible skin Paternal grandmother: GJH, Hyperextensible skin Children: GJH Grandson: GJH
884	10-19	M	9	C e, f	-	H, I u	J w	-	-	+	+	Mother: hEDS, BS Half-sister: hEDS, BS Maternal grandmother: hEDS, BS, IF Uncle: hEDS, BS
886	30-39	F	6	C -	-	H u	-	-	-	-	-	Son: hEDS, BS, GORD Daughter: hEDS, BS Mother: hEDS, BS, IF Brother: hEDS, BS
922	30-39	F	6	A, C f	E k	H, I u	-	-	-	-	-	Brother: GJH, TS, PE
967	10-19	F	8	C a, d, f, i	-	H, I s, u	-	-	-	+	-	Mother: GJH, PGMW Maternal grandmother: PGMW Maternal aunt: GJH, PGMW
1263	30-39	F	5	C d, f	D n, r	H, I u	-	-	-	-	-	-
1289	10-19	F	9	A, C a, d	D	H, I s, u	J y	-	-	-	-	Raynaud disease OMI Mother: GJH Maternal cousin: GJH
1337	40-49	F	5	C d	E	H u	-	-	Carotid artery dissection	+	-	Mother: GJH Sister: GJH, CHD
1341	30-39	F	8	C d, i	D	H s, t, u	-	-	-	-	-	Father: Shoulder subluxation Brother: Shoulder subluxation Sister: Shoulder subluxation Maternal grandfather: VV Maternal uncle: GJH, VV, MVP Maternal aunt: VV
1344	40-49	F	-	A, C a, d, h, i	-	H, I s	-	OP	-	-	-	Father: GJH
1393	0-9	F	5	C d, e, i	-	H, I s, t, u	-	-	+	-	-	Mother: JHM, HS Father: JHM, TS, marfanoid Brother: JHM, SS, Multiple maternal relatives with GJH
1397	0-9	F	5	C -	-	H u	-	-	-	-	-	Mother: hEDS Brother: hEDS
1399	30-39	F	4	C d	-	H s, u	-	-	-	+	-	Son: hEDS Daughter: hEDS

1403	40-49	M	7	C a,d	E	H,I u	J x,y	-	SaH Aor	-	-	-	Brothers: TS Maternal uncle: PE Son: PE
1421	10-19	M	7	C a	-	H,I u	-	-	-	-	-	-	Mother: hEDS Maternal grandfather: Abnormality of bladder, GJH
1422	40-49	F	-	A,C f	-	H,I u	J w	Sco	-	-	-	-	Father: Abnormality of bladder, GJH Son: hEDS
1424	0-9	F	9	A,C e	-	H,I u	-	PE	-	-	+	-	Mother: GJH Father: GJH
1425	20-29	F	-	C -	-	H -	-	-	-	-	+	+	Mother: GJH Father: GJH
1431	30-39	F	3	A,C d,f,g	-	H,I s,u	-	CHD	renal pelvis bleed	-	-	+	Father: TS, Kyph, My, RD Paternal uncle: My, RD Paternal aunt: My, RD Brother: My, RD Paternal cousin: Sudden cardiac death Paternal relative: Sudden cardiac death, GJH
1437	40-49	F	8	A,C -	E -c	H,I u	-	-	-	-	+	-	Father: GJH Son: GJH
1438	10-19	M	5	A,C f	-	H,I u	J w,y,bb	TS	-	-	Con	-	Mother: GJH, Arthralgia, Dysautonomia Brother: hEDS
1439	10-19	M	7	A,C f,g	-	H,I u	J w,bb	-	-	-	-	-	Mother: GJH, Arthralgia, Dysautonomia Brother: hEDS
1443	20-29	F	6	C d,e	-	H t	-	-	-	-	+	+	Paternal grandmother: AAA Maternal grandfather: ICA
1444	30-39	F	6	- -	- -	H -	- -	-	-	-	+	+	Cousin: GJH
1450	30-39	F	-	B,C a	-	H,I t,u,PROM	-	Str	-	-	-	-	Mother: GJH, recurrent miscarriage Sister: GJH
1455	50-59	M	6	A,C	-	H,I u	-	tall stature OP aortic ejection click	VV	-	-	-	Daughter: GJH, TS
1461	30-39	F	5	C d	- r	H t	-	-	-	-	-	+	Maternal grandfather: AAA; TS Nieces from both paternal and maternal side: GJH
1462	20-29	F	8	C a,d,f	-	H t	J w,aa	PE OP	-	-	+	+	Mother: GJH, PP, Dysautonomia Sister: Arthralgia
1464	70-79	F	-	C	-	H	-	-	-	-	-	-	-
1477	20-29	M	7	C d,i	-	H,I s,t,u	-	-	-	-	-	-	Brother: GJH
1482	50-59	F	5	A,C d	D r	H,I s,t,u	-	tall stature	-	-	-	-	Father: HTAD age 69 Mother: GJH, Raynaud disease Daughter: GJH Paternal uncle's daughter: Knee dislocation
1484	50-59	F	4	C d,h	-	H s,t,u	-	-	VV	-	-	-	Mother: VV Father: VV Sisters: VV Sons: pain susceptibility, GJH Daughter: pain susceptibility
1491	20-29	F	6	C d,f	-	H t	- y	-	-	-	-	-	-
1495	20-29	F	8	C d	-	H,I t,u	-	flexion contractures	-	-	-	+	Father: spina bifida Mother: GJH
1498	40-49	M	-	A,C l	-	H,I u	J y,bb	tall stature	-	-	-	-	Mother: GJH Daughter: hEDS
1499	10-19	F	5	A,C i	-	H,I t,u	J,L y,bb	-	-	-	+	+	Father: GJH, Sco
1500	20-29	F	4	B,C d,e,f	E	H u	-	-	SaH	-	-	-	Mother: GJH
1502	10-19	F	8	A,C d,e,f	- r	H,I s,t,u	-	umbilical hernia	Epistaxis	-	-	+	Mother: Epistaxis, GJH, PGMW Maternal aunt: Epistaxis Maternal great- grandmother: Cerebral Hemorrhage Father: TS, Hyperextensible skin Brother: GJH
1507	30-39	M	-	B,C a,f,g	-	H,I u	-	MVP TS OP Sco	-	-	-	-	Mother: GJH Sister: GJH

1511	10-19	M	6	A, C d, i	-	H, I u	-	-	-	-	-	-	Mother: GJH Maternal grandfather: GJH Brother: hEDS Sister: GJH
1526	30-39	F	3	C f, g	-	H u	-	-	-	-	-	-	Mother: VV, PGMW Brother: GJH Son: hEDS Cousins (maternal side): hEDS
1527	10-19	M	3	C d, f	-	H u	-	-	-	-	-	+	Mother: hEDS Maternal grandmother: VV, PGMW Maternal uncle: GJH
1530	10-19	F	6	- g	-	H, I u	-	tall stature	-	-	-	-	Mother: Str Father: Str, GJH Brother: Str
1579	50-59	F	6	C d, f	-	H s, t, u	-	PGMW	-	-	-	+	Father: AAA Son: -
1580	30-39	F	-	C d	-	H s, t, u	-	-	-	-	-	-	Mother: GJH
1581	40-49	F	7	C f	-	H u	-	-	-	-	-	-	-
1582	50-59	F	7	C d, e, f	-	H, I t, u	-	-	-	-	-	+	Son: hEDS
1595	10-19	F	7	C a	-	H, I u	-	-	-	-	-	-	Mother: hEDS Sister: GJH Maternal aunt: GJH
1596	50-59	F	-	C -	-	H t, u	-	-	-	-	-	+	Sister: GJH, Hyperextensible skin Daughters: hEDS
1600	20-29	F	8	C d, f	-	H t, u	-	PP Sco	-	-	-	+	Father: GJH Sister: GJH Paternal grandfather: GJH Paternal uncles: GJH Paternal cousin: GJH
1603	30-39	F	6	C f	-	H t, u	-	-	-	-	-	+	Paternal grandmother: GJH
1605	30-39	F	4	- -	-	H t	-	-	-	-	-	+	N/A
1607	40-49	F	6	C d, f	-	H, I t, u	-	-	-	-	-	+	Son: hEDS
1609	30-39	F	8	C -	-	H t	-	-	-	-	-	+	+ Crohn's disease
1613	50-59	F	5	C a, d	-	H, I s, t	-	PP	-	-	-	-	-
1616	20-29	F	7	C d	-	H, I s, t	-	PP	-	-	-	-	-
1618	30-39	F	8	C d, g	-	H t	-	-	-	-	-	-	-
1620	20-29	M	6	C d, f	-	H, I t, u	-	-	-	-	-	-	-
1626	10-19	F	8	C d	-	H u	-	-	-	-	-	+	-
1629	30-39	F	5	C d, f	n	H, I s, t, u	-	PGMW Str	-	-	-	+	Sister: hEDS Son: GJH
1630	30-39	F	8	C a, d	-	H, I t	-	-	-	-	-	+	-
1641	30-39	F	7	C d	-	H u	-	PP	-	-	-	+	-
1642	20-29	F	-	C -	-	H t	-	-	-	-	-	-	-
1656	20-29	F	7	C d, f	-	H -	-	-	-	-	-	+	-
1665	30-39	F	8	C a, d, f	-	H, I s, t, u	-	Sco	-	-	-	+	Maternal grandmother: GJH Niece: GJH
1666	10-19	F	8	C -	-	H t	-	-	-	-	-	+	-
1669	30-39	F	8	C d	-	H, I s, t	-	PP	-	-	-	+	-
1681	40-49	F	7	C a, d, f	-	H, I t	-	-	-	-	-	+	-
1682	30-39	F	8	C d	-	H t	-	-	-	-	-	+	-
1695	20-29	F	8	C f	-	H, I u	-	-	-	-	-	+	Mother: GJH
1714	40-49	F	5	C -	-	H t	-	CHD	-	-	-	+	-
1717	40-49	F	7	C d	-	H t	-	Palpitations	-	-	-	-	-
1743	20-29	F	7	C d, f	-	H, I s	-	Kyph	-	-	-	+	-

EDS Diagnostic Criteria and Abbreviations as per lists in Supplementary Table 1.