Supplementary Table S1. Clinical issues (physical features, events, complications and treatment) described in a single patient with mcEDS-*CHST14*

Events in deliveries	Denuded skin of the cheek during delivery, preterm delivery at 32
	weeks of gestation, vacuum extraction, premature rupture of
	membranes
Events in neonatal	Hypoglycemia, admission to the neonatal intensive care unit, small
periods	for gestational age
Craniofacial features	Premature sagittal synostosis, large skull, triangular face, anterior
	hair whorl, low posterior hairline, epicanthus, deep-set eyes, puffy
	eyelids, upturned nose, shallow palate, bifid uvula, overbite,
	hypoplastic maxilla
Skeletal features	Carpal tunnel syndrome, dural ectasia, elbow bursitis, femoral
	anteversion, C5-6 luxation after falling, incomplete ossification of
	the C1 ring, unilateral vertebral fusion at T3-4, spondylolisthesis
Cutaneous features	Sacral skin tag, ganglion of the foot, gray scalp hair, puffy lower
	limbs and feet, chronic ulcers, wrinkling skin (abdominal wall,
	dorsal hand/foot), sagging skin on the face and chest, cysts (buttock
	and knee), fetal finger pads, subcutaneous calcified spheroids,
	severe eczema, localized burning sensation of the skin, androgenic
	alopecia (woman), hyperpigmentation
Cardiovascular or	Catheter-related large thrombus, infective endocarditis, epidural
related	bleeding associated with obstructive hydrocephalus, subdural
complications	hematoma, postural orthostatic tachycardia syndrome, polymorphic
	ventricular extrasystoles
Gastrointestinal	Hiatal hernia with gastroesophageal reflux, duodenal obstruction
complications	due to malrotation, abdominal bloating after eating, lower
	gastrointestinal tract hemorrhage, infralevator anal atresia,
	cholelithiasis
Genitourinary	Horseshoe kidney, atresia of vas deferens, gross hematuria, double
abnormalities	ureter and collecting system, hypoplastic labia majora
Ophthalmologic	Phthisis bulbi, irregular nasally displaced pupil, conjunctival
abnormalities	injection, prominent bulging eyes, nasolacrimal duct stenosis,
	nocturnal lagophthalmos, Peter's anomaly, nystagmus
Otologic	Narrow middle ear space, sensitivity to loud noise, cerumen
abnormalities	impaction
Structural brain	Gray matter heterotopias, hydrocephalus, hippocampal hypoplasia,
anomalies	- by the first of the state of

Neurological	Brain contusion after falling, reduced deep tendon reflex, cerebral
features	venous thrombosis, dysarthria, intraventricular hemorrhage,
	migraine, radicular pain, subarachnoid hemorrhage, syringomyelia
Muscular features	Spontaneous rupture of abdominal muscles, thin and high
	diaphragm
Surgical indication	Carpal tunnel syndrome, cataract, cleft lip/palate, duodenal
	obstruction due to malrotation, epiblepharon, epidural hematoma
	with obstructive hydrocephalus, exudative otitis media, femoral
	anteversion, fistula, cholelithiasis, hydrocephalus, infective
	endocarditis, low imperforate anus, massive skin necrosis,
	protruding coccyx, ptosis, sacral abscess, spinal hematoma,
	subcutaneous cyst, tethered spinal cord, tibial tuberosity fracture
Pharmacologic	Medication for anxiety, asthma, depression, epilepsy,
therapy	hyperlipidemia, hyperuricemia