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

0 · F
Denuded skin of the cheek during delivery, preterm delivery at 32
weeks of gestation, vacuum extraction, premature rupture of
membranes
Hypoglycemia, admission to the neonatal intensive care unit, small
for gestational age
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
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
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
Catheter-related large thrombus, infective endocarditis, epidural
bleeding associated with obstructive hydrocephalus, subdural
hematoma, postural orthostatic tachycardia syndrome, polymorphic
ventricular extrasystoles
Hiatal hernia with gastroesophageal reflux, duodenal obstruction
due to malrotation, abdominal bloating after eating, lower
gastrointestinal tract hemorrhage, infralevator anal atresia,
cholelithiasis
Horseshoe kidney, atresia of vas deferens, gross hematuria, double
ureter and collecting system, hypoplastic labia majora
Phthisis bulbi, irregular nasally displaced pupil, conjunctival
injection, prominent bulging eyes, nasolacrimal duct stenosis,
nocturnal lagophthalmos, Peter's anomaly, nystagmus
Narrow middle ear space, sensitivity to loud noise, cerumen
impaction
Gray matter heterotopias, hydrocephalus, hippocampal hypoplasia,
Gray matter neterotopias, nyarocepharas, inppoeampar nypopiasia,

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