

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 periods	Hypoglycemia, admission to the neonatal intensive care unit, small 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 related complications	Catheter-related large thrombus, infective endocarditis, epidural bleeding associated with obstructive hydrocephalus, subdural hematoma, postural orthostatic tachycardia syndrome, polymorphic ventricular extrasystoles
Gastrointestinal complications	Hiatal hernia with gastroesophageal reflux, duodenal obstruction due to malrotation, abdominal bloating after eating, lower gastrointestinal tract hemorrhage, infralevator anal atresia, cholelithiasis
Genitourinary abnormalities	Horseshoe kidney, atresia of vas deferens, gross hematuria, double ureter and collecting system, hypoplastic labia majora
Ophthalmologic abnormalities	Phthisis bulbi, irregular nasally displaced pupil, conjunctival injection, prominent bulging eyes, nasolacrimal duct stenosis, nocturnal lagophthalmos, Peter's anomaly, nystagmus
Otologic abnormalities	Narrow middle ear space, sensitivity to loud noise, cerumen impaction
Structural brain anomalies	Gray matter heterotopias, hydrocephalus, hippocampal hypoplasia, optic nerve hypoplasia, pachygyria

Neurological features	Brain contusion after falling, reduced deep tendon reflex, cerebral 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 therapy	Medication for anxiety, asthma, depression, epilepsy, hyperlipidemia, hyperuricemia