

Table 1 supplement: Clinical features of affected cases

	UP IV:2	UP IV:3
Intelligence	Normal	Normal
Height	Short Stature (152 cm)	Short Stature (159 cm)
Head	-	Brachycephaly
	Curly hair	Curly hair
Neck	Webbed neck	Webbed neck
	Low posterior hairline	Low posterior hairline
Face	Facial asymmetry	Facial asymmetry
	Decreased facial expression	Decreased facial expression
	Mild micrognathia	Mild micrognathia
Eyes	Ophthalmoplegia*	Ophthalmoplegia*
	Down-slanting palpebral fissures	Down-slanting palpebral fissures
	Epicanthal folds (right)	-
	Ptosis (right)	-
	Compound myopic astigmatism OD: -3.0 -4.0 X 180, OS: -2.0 -4.50 X 180	Compound myopic astigmatism OD: -4.75 -3.75 X 75, OS: -2.5 -7.25 X 80
	Arched eyebrows	Arched eyebrows
Nose	Prominent nasolabial folds	Prominent nasolabial folds
	Bulbous nose	Bulbous nose
Mouth	Small mouth	Small mouth
	Small oral opening	Small oral opening
	High arched palate	High arched palate
	-	Irregular teeth
Ears	-	Posteriorly angulated ears
Spine	Neck contracture	Neck contracture
	Scoliosis (left)	Scoliosis (left)
	Scapular winging	Mild Scapular winging
Thorax	Thoracic hypokyphosis	Thoracic hypokyphosis
Limbs	Patellar hypoplasia (bilaterally)	Patellar hypoplasia (left)
	Limited flexion of knee	Limited flexion of knee
	Limited abduction of left shoulder	-
Hands	Limited abduction of wrists	Limited extension of wrists

	Camptodactyly	Camptodactyly
Feet	Pes cavus (right)	Pes cavus
	-	Hallux valgus
Muscle	Decreased muscle mass (lower limbs)	Decreased muscle mass (lower limbs)

Investigations

Hand X-ray	Coalition between scaphoid and hamate	No fusion
MR biopsy	Fibrosis	Fibrosis
Brain and orbital MRI	Normal	Normal
EMG	Normal	Normal
Nerve conduction	Normal	Normal
ERG	Normal	Normal

MR denotes medial rectus. MRI denotes magnetic resonance imaging. EMG denotes electromyography. ERG denotes electroretinography.

*IV:2 underwent medial rectus muscle resection (7mm) and lateral rectus recession (6mm). IV:3 underwent bilateral lateral rectus recessions (11mm).