		Patient's Initials (N.S.)
	Referring physician name	
	Referring doctor email	
	Date of birth	
	Gender	
	Age at last examination	
	Country	
	Molecular data	
	KMT2A (MLL) nucleotide alteration	
	KMT2A (MLL) amino-acid alteration	
	Other gene variants or CNVs	
	Clinical data	
Growth Parameters	Intra-uterine growth retardation	
	Birth weight (°p)	
	Birth lenght (°p)	
	Birth CC (°p)	
	Failure to thrive	
	Weight at last examination (°p)	
	Height at last examination (°p)	
	CC at last examination (°p)	
	SPAN/stature at last examination	
Neuropsychiatric signs	Developmental delay	
	Intellectual disability (mild/moderate/severe)	
	Speech delay	
	Wide-base gait	
	Hypotonia	
	Seizures	
	Aggressive behaviour	
	Autistic features	
Cr ani	Round and Flat face	

	Tick eybrows	
	Long eyelashes	
	Hypertelorism	
	Downslanting palpebral fissures	
	Vertically Narrow palpebral fissures	
	Low-set ears	
	Wide nasal bridge	
	Broad nasal tip	
	Thin vermillion border	
	High-arched palate	
	Hypodontia/Abnormal dentition	
Skin and hair dysmorphisms	Thick hair	
	Hypertichosis (generalized/localized)	
	Hypertichosis cubiti	
Skeletal abnormalities	Delayed bone age	
	5th finger clinodactily	
	Short/Thick limbs	
	Sacral dimple	
	Short fingers	
	C2-C3 vertebral fusion	
	Other skeletal anomalies	
Clincial findings	Feeding problems	
	PEG or NG feeding	
	Costipation	
	Ocular abnormalities	
	Strabismus	

	Recurrent urinary tract infections	
	Immunodeficiency	
	Kidney/ureter malformation	
	Cardiac anomalies	
	Other internal organ problems	
	Other clinical signs	
Instrumental findings	**Brain MRI and spinal cord MRI**	
	Foramen magnum stenosis	
	Skeletal X-Ray	
	Cervical CT scan	
	Surgical intervention	