

Table S1. Clinical features of WBS patients. NA, not available. “+” stands for present; “-” stands for not present. A more exhaustive description of neuropsychological testing was reported in Table 1.

Clinical features		WBS154	WBS166	WBS160	WBS179
Intellectual Disability		Moderate	Moderate	Severe	Severe
Cardiovascular	Supravalvular aortic stenosis	-	+	-	-
	Peripheral pulmonary stenosis	-	+	-	+
	Valvular pulmonic stenosis	-	-	-	+
	Hypertension	-	+	-	-
	Others	-	-	-	-
Craniofacial	Wide mouth	-	-	+	+
	Prominent ear lobes	-	-	-	-
	Dolicocephaly	-	-	-	-
	Broad forehead	-	-	-	+
	Microcephaly	+	-	+	+
	Bitemporal narrowing	+	-	+	-
	Periorbital fullness	+	+	+	+
	Epicanthal folds	-	+	+	+
	Stellate irides	-	-	+	-
	Malar flattening	+	+	+	+
	Short upturned nose	-	-	+	+
	Bulbous nasal tip	-	-	-	-
	Bridge nose flattened	-	-	-	-
	Long philtrum	+	-	+	+
	Full lips	+	-	+	+
	Full cheeks	+	+	+	+
	Dental abnormalities/malocclusion	-	-	+	+
Endocrine	Subclinical hypothyroidism	-	-	-	-
	Precocious puberty	-	NA	-	NA
	Hypercalcemia	-	-	-	-
	Glucose intolerance or diabetes mellitus	-	-	-	-
Gastrointestinal	Feeding difficulties	-	-	+	+
	Abnormal weight gain	-	-	+	+
	Celiac disease	-	-	-	-
	Constipation	-	+	-	+
	Gastroesophageal reflux	-	+	+	+
	Abdominal pain (unclear cause)	-	-	-	-
	Rectal prolapse	-	-	-	-
Genitourinary	Congenital malformation	-	-	-	-
	Enuresis	-	-	-	-
Ocular	Strabismus	+	+	+	+
	Hypermetropia	-	-	-	-
Musculoskeletal	Kyphosis	+	-	+	+
	Hyperlordosis	-	-	+	-
	Radioulnar synostosis	-	-	-	-
	Umbilical hernia	-	-	-	+
Other	Hoarse voice	-	+	+	+
	Hyperacusis	+	+	+	+
	Epilepsy	-	-	+	-
	Scoliosis	-	-	+	-

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Eliminato: +