

資料 4. 全国疫学調査結果：日本人 SYS 患者の症状

	Our study		McCarthy et al.[2]	
Neonatal hypotonia	23/24	96%	66/68	97%
Poor suck in infancy	19/23	82%	62/64	97%
Developmental delay	25/25	100%	70/70	100%
Distal joint contractures	21/25	83%	69/78	88%
Autism spectrum disorder	10/20	50%	25/32	78%
Characteristic facial features	25/25	100%	21/26	81%
Sleep apnea	11/21	52%	38/50	76%
Short stature	19/24	79%	12/20	60%
GH deficiency	11/13	85%		
Temperature instability	9/20	45%	40/60	67%
Respiratory distress	14/25	56%	41/58	71%
Chronic constipation	10/23	43%	39/55	71%
GERD	6/20	30%	34/60	57%
Scoliosis	17/24	71%	26/46	57%
Eye abnormalities	23/25	92%	14/18	78%
Hypogonadism	2/8	25%	26/64	41%
Genital hypoplasia	3/24	21%		
Small hands	23/25	92%	13/18	78%
Hyperphagia	3/22	14%	14/56	25%
Sleep disturbance	10/20	50%		
Characteristic behavior	5/18	28%		
Episode of encephalopathy	4/23	17%		