

Clinical criteria	Points	Patient reported here
Cerebriform connective tissue nevus	5	-
Asymmetric, disproportionate overgrowth (one or more) (a) Limbs or (b) Hyperostosis of the skull or (c) Hyperostosis of the external auditory canal or (d) Megaspondylodysplasia, scoliosis, or rib hyperostosis	5	5
Organ/visceral overgrowth (two or more) (a) Central nervous system or (b) Urogenital system or (c) Eye or (d) Spleen or (e) Kidney or (f) Liver or (g) Tonsils or adenoids or (h) Gingiva or tongue	5	5
Bullae or cysts of the lungs	2	-
Dysregulated adipose tissue (one or more) (a) Lipomas or (b) Lipodystrophy or (c) Myocardial septal lipoma	2	2
Linear verrucous epidermal nevus	2	-
Vascular malformations (one or more) (a) Capillary malformation or (b) Venous malformation or (c) Lymphatic malformation	2	-
Specific tumors (a) Female genitourinary cystadenoma (<11 yo) or (b) Parotid monomorphic adenoma (<11 yo) or (c) Meningioma (meningotheial and transitional subtype) or (d) Testicular cystadenomas or cystadenocarcinomas	1	-
Facial phenotype (three or more features) (a) Dolichocephaly (b) Long face (c) Down slanting palpebral fissures and/or minor ptosis (d) Low nasal bridge (e) Wide or anteverted nares (f) Open mouth at rest	2	-
Deep vein thrombosis and/or pulmonary embolism	2	-
Substantial prenatal extracranial overgrowth	-5	-
Ballooning overgrowth	-5	-
Score		12

Table S1 Clinical data of the patient described in the present report based on the dyadic genotype–phenotype score system for the diagnosis of Proteus syndrome suggested by Sapp et al. (2019). A score of ≥ 10 points with a mosaic *AKT1* variant or ≥ 15 points with or without an *AKT1* mosaic variant. In light of the identified *AKT1* variant, Proteus syndrome can be diagnosed in the patient described here.