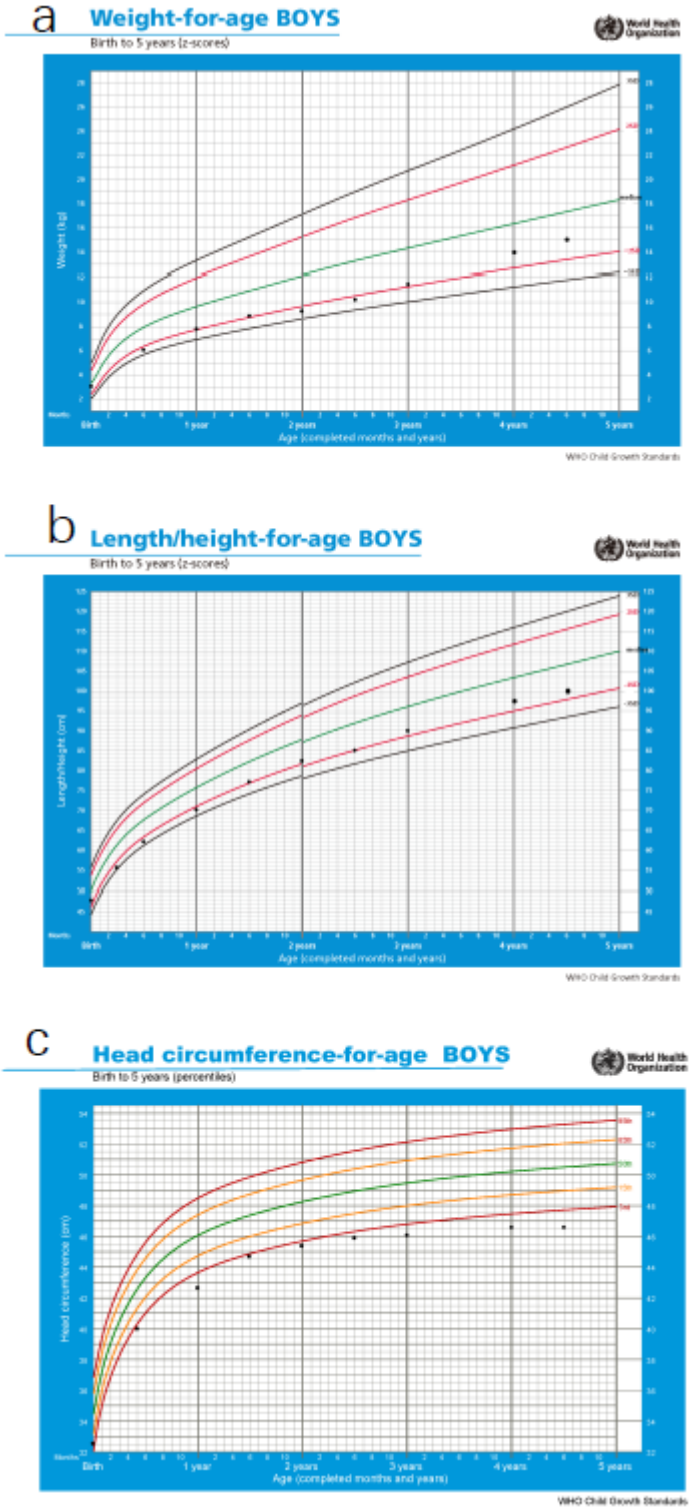

Supplemetary Figure 1 Growth Charts of the proband.(a) weight for age chart (b)length for age chart (c) head circumference for age chart

Supplementary Table 1 Clinical information on our patient and reported BRPS patients

		Our patient	Frequency in reported cases[1-25]
Sex		M	24F, 23M, 2NA
SGA		-	6/43 (14.0%)
Growth	Failure to thrive	+	33/43 (76.7%)
	Poor growth	+	35/44 (79.5%)
	Short stature < P3#	-	17/40 (42.5%)
Head	Microcephaly < P3#	+	26/43 (60.5%)
	Trigonocephaly	-	3/43 (7.0%)
Facial Dysmorphism[a]		+	33/44 (75%)
Face	Prominent forehead	+	19/44 (43.2%)
	Micrognathia	-	10/44 (22.7%)
Eyes	Hypertelorism	+	12/44 (27.3%)
	Downslanting palpebral fissures	+	22/44 (50.0%)
	Strabismus	+	19/43 (44.2%)
	Arched eyebrows	+	18/32 (56.3%)
Nose	Prominent nasal bridge	-	7/44 (15.9%)
	Short nose	-	7/44 (15.9%)
	Anteverted nares	-	17/32 (53.1%)
Mouth	High-arched palate	-	25/44 (56.8%)
	Crowded teeth	+	5/36 (13.9%)
Ears	Low-set ears	+	9/26 (34.6%)
Gastrointestinal	Feeding difficulties	+	38/43 (88.4%)
Skeletal	Scoliosis	-	6/37 (16.2%)
	Arachnodactyly	-	9/35 (25.7%)
	Ulnar deviation of hands at rest	-	13/35 (37.1%)
Muscle	Hypertonic extremities	+	13/43 (30.2%)
	Generalized or trunk hypotonia	+	41/45 (91.1%)
Central Nervous System			
	Developmental delay	+	47/47 (100.0%)
	Intellectual disability	+	43/43 (100.0%)
	Poor or absent speech	+	42/42 (100.0%)
	Seizures	-	15/46 (32.6%)
Behavioral Manifestations			
	Autistic features	+	20/31 (64.5%)

Notes: +: positive, -: negative, [a]: patients with equal or greater than 3 facial

characteristics were defined as facial dysmorphism; #: the value was compared with WHO child growth standards.

Abbreviation: F: female, M: male, NA: not available.

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四川大学华西第二医院医学伦理委员会

伦理备案批件

医学科研 2018 伦审批第（019）号

课题负责人：李晋蓉，研究项目《中国西部儿童生长发育障碍性疾病遗传学研究及治疗随访》。本研究已提交研究方案、临床研究知情同意书、本院研究人员简历等材料，研究对象均在研究前签署知情告知。经四川大学华西第二医院医学伦理委员会审议，同意该项目备案。

四川大学华西第二医院医学伦理委员会

日期：2018 年 6 月 30 日

备案材料名称：

1、 医学伦理备案申请表；

2、 研究方案；

3、 知情同意书；

4、 本院研究人员简历。