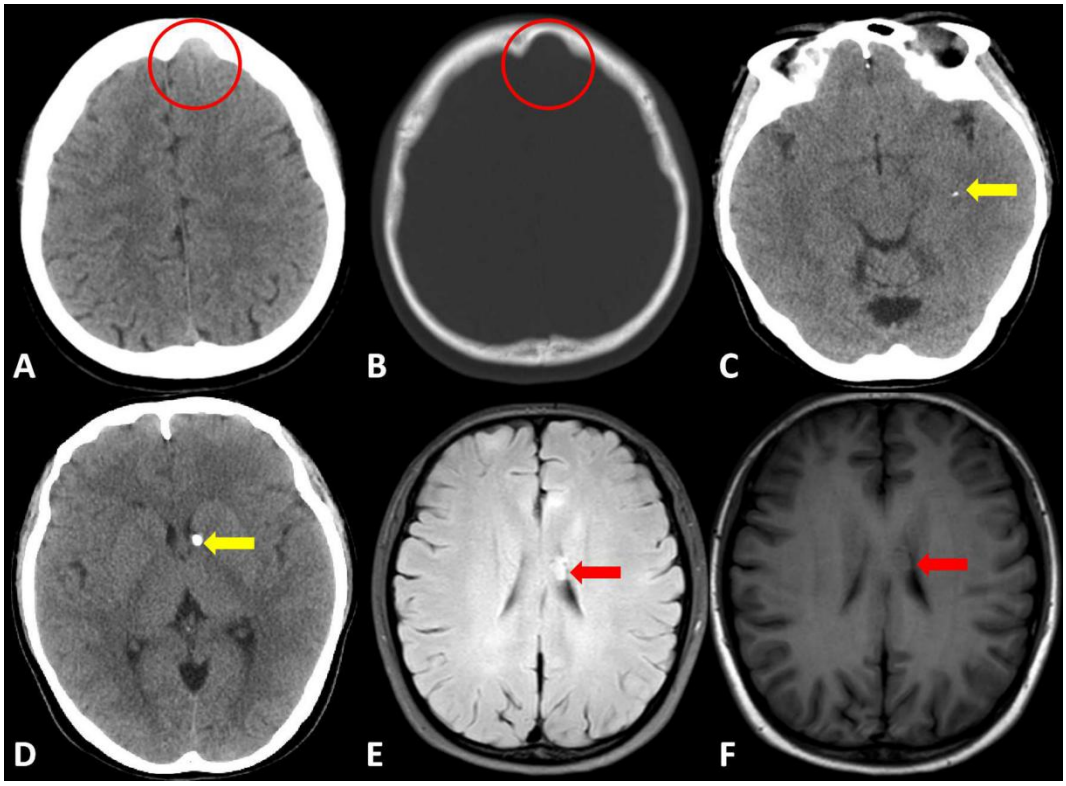
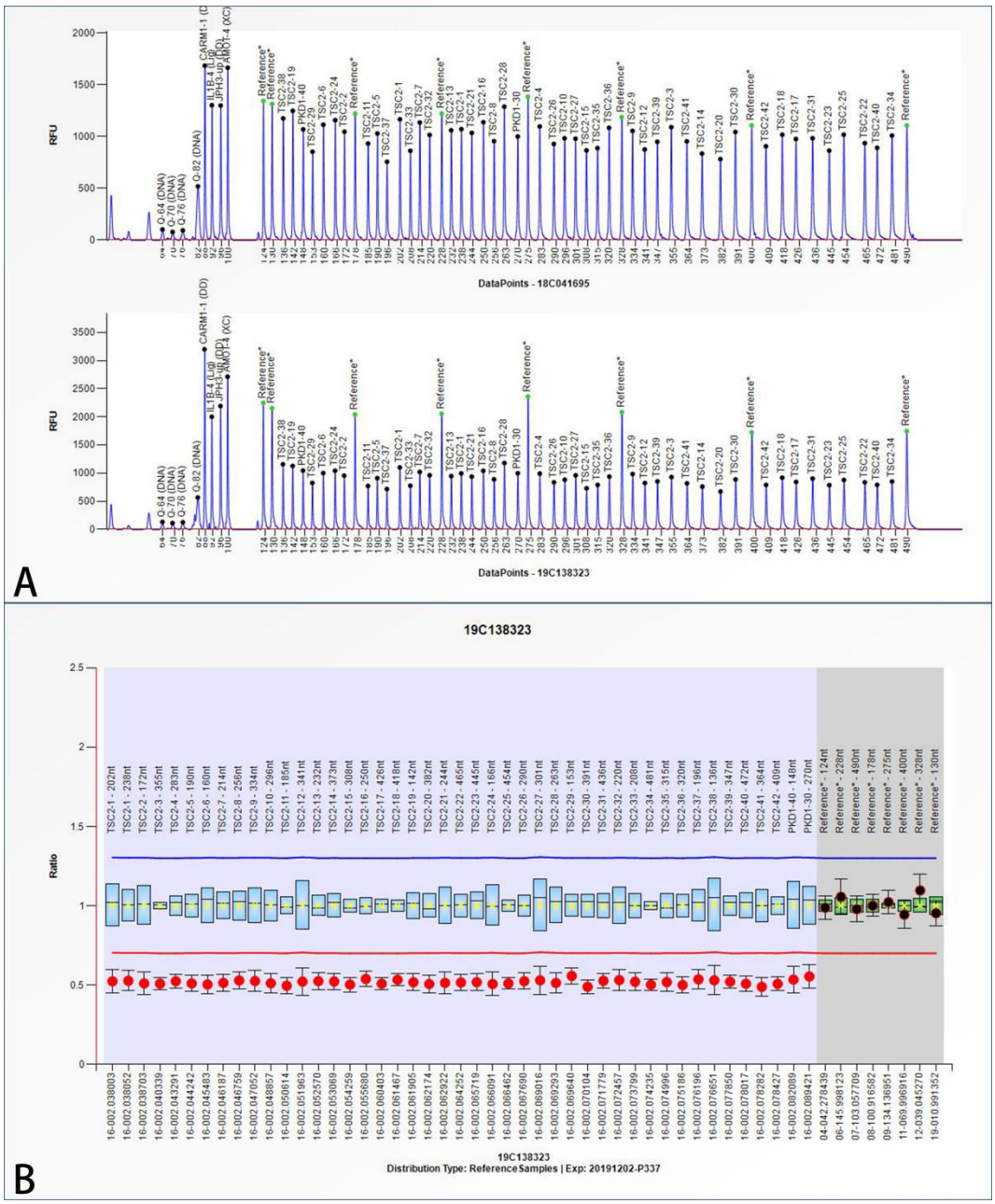
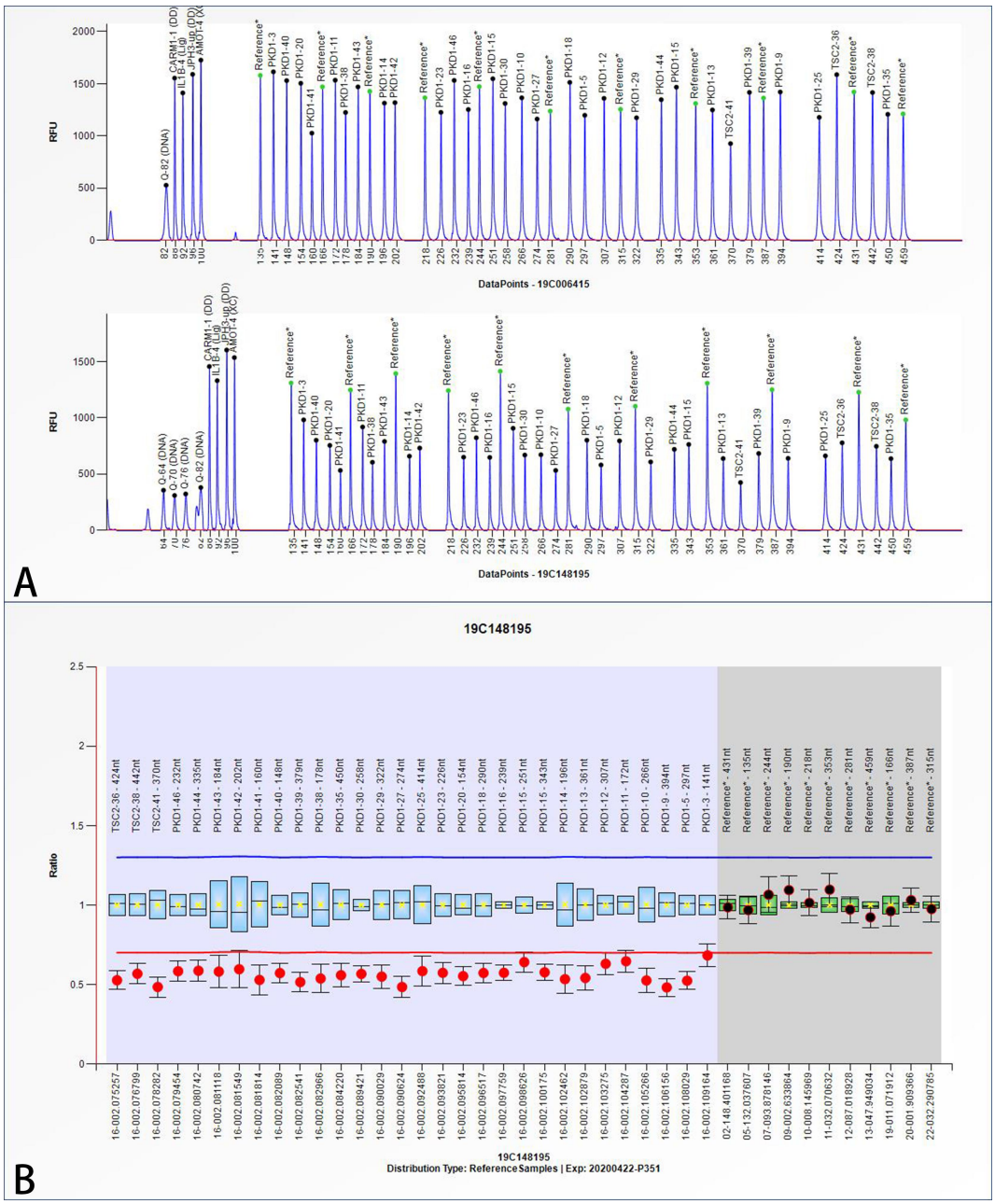


Supplementary Figure S1: Cranial computed tomography (CT) and magnetic resonance imaging (MRI). Cranial CT demonstrated (A-B). a slightly high density nodular shadow with a clear boundary in the left temporal lobe which measured 14 × 10mm (red circles) and (C-D). multiple calcified nodules (yellow arrows). (E-F). Brain MRI demonstrated tulbercles and calcified nodules in the left lateral ventricle and subcortical demyelination (red arrows).



Supplementary Figure S2: Using multiplex ligation-dependent probe amplification to do TSC2 genetic detection (Normal control: 18C041695, Patient sample: 19C138323). (A). Compared with normal control, patient's output indicates the deletions occur in whole *TSC2* and extend into neighbouring *PKD1*. (B). The dots present fluorescence intensity. Black dots between 0.7 and 1.3 present normal condition. Red dots show reductions of exon 1-42 in the *TSC2* gene and exon 30 and 40 in the *PKD1* gene by half of genomic quantity more intuitively, it means heterozygous deletions.



Supplementary Figure S3: Using multiplex ligation-dependent probe amplification to do PKD1 genetic detection (Normal control: 19C006415, Patient sample: 19C148195). (A). Compared with normal control, patient's output indicates the deletions occur in whole *PKD1* and neighbouring *TSC2*. (B). Red dots present heterozygous deletions of *TSC2* upstream (exon 36,38,41) and *PKD1* (exon 3,5-7,9-16,18,-23,25-27,29-31,33,35-44,46). The absence of so many exons is generally continuous, which can almost present deletion of whole *PKD1*.

Supplementary Table S1: Diagnostic criteria for TSC.

* Includes tubers and cerebral white matter radial migration lines.

Major features	1. Hypomelanotic macules (≥3, at least 5 mm diameter) 2. Angiofibromas (≥3) or fibrous cephalic plaque 3. Ungual fibromas (≥2) 4. Shagreen patch 5. Multiple retinal hamartomas	6. Cortical dysplasias* 7. Subependymal nodules 8. Subependymal giant cell astrocytoma 9. Cardiac rhabdomyoma 10. Lymphangioleiomyomatosis (LAM) 11. Angiomyolipomas (≥2)
Minor features	1. "Confetti" skin lesions 2. Dental enamel pits (≥3) 3. Intraoral fibromas (≥2)	4. Retinal achromic patch 5. Multiple renal cysts 6. Nonrenal hamartomas
Genetic detection	1. *TSC1* pathogenic mutation	2. *TSC2* pathogenic mutation
Definite diagnosis	I. Two major features (exclude only exists major feature 10 and 11) II. One major feature with two or more than two minor features III. The identification of either a *TSC1* or *TSC2* pathogenic mutation	
Possible diagnosis	I. One major feature II. One major feature and one minor feature III. Two or more than two minor features	

Supplementary Table S2: Differential diagnosis of Tuberous sclerosis complex (TSC) and Polycystic kidney disease (PKD).

Differential points	Tuberous sclerosis complex (TSC)	Polycystic kidney disease (PKD)
Family history	Frequently (Autosomal dominant inheritance, may sporadic)	Frequently (Autosomal dominant inheritance, recessive dominant inheritance, may sporadic)
First diagnosis	Usually children	Usually adults (ADPKD) Usually children (ARPKD)
Typical features	Triad syndrome includes skin lesions, mental retardation and seizure.	None
Renal damages	PKD usually combines with renal angiomyolipomas (HU is inhomogeneous: high-low density, irregular calcification)	Only PKD (HU is homogeneous: low density, annular calcification)
Extra renal damages	May have multiple organ damages, for example skin, brain, heart, lung and etc (Supplementary Table 1).	May have multiple organ damages, for example cysts of liver, pancreas, seminal vesicle, spleen and arachnoid, extracranial aneurysm and etc. (ADPKD) May have congenital hepatic fibrosis. (ARPKD)
Genetic detection	*TSC1* or *TSC2* pathogenic mutation	*PKD1* or *PKD2* pathogenic mutation