Introduction
Tuberous sclerosis complex (TSC) is a hereditary disease that caused by \textit{TSC1} and \textit{TSC2} mutation. The main pathological changes are hamartoma and tissue constitution deficiency, which can involve many organs and tissues.

Methods
Ninety-six patients with TSC and their parents underwent TSC target sequencing and multiplex ligation-dependent probe amplification. All gene results were analysed for clinical phenotype and gene pathogenicity following standard criteria.

Results
Ninety-two mutations in \textit{TSC1} and \textit{TSC2} were found in 96 patients. All 26 patients with novel mutations were clinically diagnosed with TSC and had typical clinical phenotypes. Among them, 22 were point mutations, and four were large duplications/deletions; 18 were sporadic mutations, and eight were familial mutations; five mutations were located in \textit{TSC1}, and 21 mutations were located in \textit{TSC2}. Five were not born yet or were too young to be assessed for intelligence. The proportion of patients with intellectual disability was 50\% (3/6) for those with frameshift mutations, 71\% (5/7) for those with nonsense mutations and 50\% (1/2) for those with splicing mutations. The incidence of intellectual disability was 10/18 in patients with point mutations and 3/3 in those with large duplications/deletions (P < 0.05).

Conclusion
This study extends our knowledge of the spectrum of \textit{TSC1} and \textit{TSC2} mutations, and confirmed the patients’ clinical diagnoses based on gene analysis.