# Association between 1p36 copy number variations, abnormal expression of PEX10 gene and epilepsy phenotype

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## Objective

To investigate the association between PEX10 gene and 1p36 copy number variations concurrent epilepsy by analyzing 3 cases of copy number variants in 1p36 region.

## Methods

The karyotypes of 3 patients were determined by the application of high resolution chromosome banding, multiple connection dependence probe amplification (MLPA), fluorescence in situ hybridization (FISH) combined with single nucleotide polymorphism array (SNP) technology. Real-time PCR technique was carried out to detect mRNA expression levels of PEX10 gene in peripheral blood of the patients.

## Results

No abnormality was present by high resolution karyotype analysis. MLPA analysis showed that all the 3 patients had a copy number variation of the subtelomeric region in one short arm of chromosome 1, which was confirmed by FISH analysis and SNP chips. Case 1 and case 2 both had epilepsy phenotype and their copy number variations covered PEX10 gene. On the other hand, case 3 didn't have epilepsy, and the PEX10 gene copy number is normal. Family pedigree investigation showed all the chromosome abnormality of the 3 cases were de novo. Compared with the ordinary people, the real time-PCR showed that mRNA expression level of PEX10 gene was increased in case 1 but decreased in case 2.

## Conclusion

The abnormal expression of PEX10 gene resulting from the copy number variations of 1p36 region might be associated with the epilepsy phenotype.