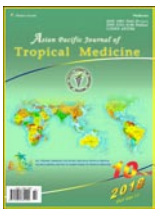

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A study on the stable inheritance of chromosome aberration karyotype in three families of two generations in Hainan province

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ABSTRACT **Objective:** To examine chromosomes of a total of 10 857 people who asked for prenatal genetic counseling in our hospital from February 1994 to July 2018. The important diagnostic reference index for the clinical diagnosis, screening for genetic abnormality and carrying out appropriate guidance have been done for them to adopt appropriate prenatal strategies and realize a first-degree prevention and control of birth defects. **Methods:** Routine G banding analysis was performed on genetic counselors. C banding, N banding analysis and whole genome sequencing were performed according to abnormal karyotypes. **Results:** The chromosome aberration karyotype 46, X, inv(Y)(p11.2q11.2) pat/45, X pat, number: 3 574 was found and reported for the first time across the globe; Besides, this aberrant chromosome karyotype experienced stable inheritance of two generations in 8 adult males of three families. **Conclusions:** The adult males in three families have normal phenotype and they can still have normal fertility. Among those people, no pathogenic gene has been detected and no genetic material has lost or increased. The same aberration karyotype has been found in all 8 adult males of two generations. The ratio of these two generations' karyotype is one out of ten thousand. As a rare genetic polymorphism, the abnormal karyotype can be regarded as an evolutionary marker, and play a unique role in paternity testing. In addition, the inverted Y chromosome can also be used to study the route of population migration. As a marker of genetic relationship, it may provide a method to study population genetics and historical geography.

Keywords: Inverted Y chromosome; Chimera; Prenatal genetic counseling

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