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Study on The Pedigrees of Three Cases of Whole-Arm Translocation in Hainan China and Literature Review: A Retrospective Study

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Abstract

In this study, in order to promote chromosome abnormality carriers eugenics, three patients with adverse pregnancy histories were examined by cytogenetics and their pedigrees further analyzed. In this retrospective study, approximately anticoagulant peripheral venous blood from the patients was collected for peripheral blood cell culture and chromosome analysis. Karyotypes were analyzed in the BEIONMED karyotype analysis system. The karyotypes of the three probands were all whole-arm translocations (WATs): case 1 (DatabaseNo.3591): 46, XY, t (7; 13) (p10; p10) dn, two years of marriage in which the spouse did not have pregnancy, with azoospermia; case 2 (Database No.3809): 46, XY, t(12; 17) (p10; q10), three spontaneous abortions within three years of marriage; case 3 (Database No.4914) 46, XX, t(2;6) (p10; q10) mat, 21ps+pat, a year of marriage without pregnancy. When the parents are carriers of WAT, the family should be considered to have a high reproductive risk, increasing the risk of producing offspring with chromosomal abnormalities. Three kinds of human chromosomal aberration karyotypes were reported for the first time providing an important basis for studying the occurrence and clinical consultation of chromosomal diseases.

Keywords: Case Report, Chromosomes, Genetic Counseling, Whole-Arm Translocation

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Introduction

Variations in the number and structure of chromosomes are linked to a variety of congenital abnormalities, intellectual disabilities and cancers. Chromosomal translocation can come from two sources: one is transmitted from the father or the mother-while the other is chromosomal aberrations occuring during gamete formation or zygote division. Because the breakage points of the two chromosomes are close to the centromere, the whole-arms will exchange when the chromosomes are translocated with each other, this process called whole-arm translocations (WATs). However, during the meiosis of germ cells, the translocated chromosomes can be paired to a quadriradial chromosome which is formed, and the related chromosomes can undergo para-location separation, ortho-1 separation, ortho-2 separation and 3:1 separation, resulting in 18 kinds of gametes. Among them, only one of them is normal; one is the carrier of balanced translocation; and the others are partial trisomy or monomer gametes. These gametes can cause abortion, stillbirth and birth defects, when combined with normal gametes (1).

Due to the same amount of genetic material, the phenotype and intelligence of patients with whole-arm chromosomal translocation can be normal. However, the genetic effect on the offspring of the translocation carriers will be obvious when they marry a normal person. Some studies (2, 3) have shown that when the parents are such carriers, the special chromosome rearrangement can affect the fertility of male offspring. Even though the composition of derivatives is unknown in two thirds of the cases, the circulation rate appears to be higher than that of other mutual translocations, possibly indicating that there are other factors, such as specific genomic polymorphisms. It has been reported (4) that hereditary WATs often appears in various tumors as unbalanced secondary changes, possibly caused by DNA repair errors. In this study, three patients with adverse pregnancy histories were examined by cytogenetics and their pedigrees further analyzed.

Materials and Methods

In this retrospective study, three cases with an adverse

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pregnancy history were examined by cytogenetics and also their pedigrees were further analyzed (Table 1). The karyotypes of the three cases were identified by the expert group of the Chinese Human Chromosome Abnormality Karyotype Database and no related reports were found by checking the Cytogenetics Database and the Chinese Human Chromosome Abnormality Karyotype Database. Therefore, the karyotype was included in the Chinese Human Chromosome Abnormal Nuclei Database. The patients provided informed consent for this study.

Table 1: Comparison among the three cases in family history of this study

Item	Case 1	Case 2	Case 3
Area	Wenchang City, Hainan Province	Qionghai City, Hainan Province	Haikou City, Hainan Province
Age (Y)	34	31	29
Sex	M	M	F
Nation	Han	Han	Han
Occupation	Company employee	Company employee	Company employee
Height (cm)	172	170	160
Visit time	2013	2014	2016
Reason	Married for two years; spouse had no pregnancy	Three spontaneous abortions within three years of marriage	PGD failed after a year of marriage without pregnancy
Pregnancy history of self or spouse	G0P0	G3P0	G0P0
Proband's karyotype	46,XY,t(7;13)(p10;p10)dn		
Database No.3591	46,XY,t(12;17)(p10;q10) Database No.3809	46,XX,t(2;6)(p10;q10)mat, 21ps+ patDatabaseNo.4914	
Spouse's karyotype	46, XX	46, XX	46, XY
Spouse's age (Y)	31	24	30

M; Male and F; Female.

Human peripheral blood lymphocyte culture medium (Guangzhou Baiyunshan Baidi Biomedical Co., Ltd.), low-osmosis solution (0.075 mol/LKCl), 10 μg/mL colchicine, fixative solution (methanol and glacial acetic acid), phosphate buffer solution, trypsin and Jimsa dye solution.

Karyotype analysis of peripheral blood lymphocyte culture

Approximately 3 mL of the peripheral patients blood was extracted to heparin sodium tubes for anticoagulation and evenly mixed. Approximately 1.5 mL of the blood was inoculated in the peripheral blood lymphocyte medium and evenly mixed. The cells were incubated in an incubator at 37°C for 72 hours, and colchicine was added to stop cell division. Then, the cells were harvested and microscope slides prepared for banding and analysis. Under an Olympus CX21 microscope, 30 cells in metakinesis were counted. Besides, 3-5 karyotypes were analyzed in the BEIONMED karyotype analysis system. For the patients with abnormal karyotypes, the modal number of cells was counted, and karyotype analysis conducted. The karyotype description referred to the International System for Human Cytogenetic Nomenclature (2016). In this study, karyotype analysis was also performed on the spouses and some family members of the probands. The pedigree chart was produced in PowerPoint (PPT).

Ethics approval and consent to participate

The patients provided informed consent to participate in this study, and their pedigrees were further investigated. This study protocol was approved by the Ethics Committee of Haikou Hospital of Traditional Chinese Medicine (HKSZYYYLL-2022(S)-08).

Results

Case 1: The proband was a 34-year-old man of Han nationality living in Wenchang City, (Hainan Province, China). The patient was a company employee, his height was 172-cm. He had a normal phenotype. Two Years of marriage without pregnancy. In September 2013, he was admitted to the Second Affiliated Medical Reproductive Center of Hainan Medical University. Karyotype(DatabaseNo.3591):46,XY,t(7;13)(p10;p10)dn (Fig.1A). Testicular biopsy indicated azoospermia. For the five items of coagulation, fibrinogen (FIB) was 1.88g/L (reference range: 2-4 g/L), while the rest was normal. The six sex hormones, including follicle-stimulating hormone (FSH), luteinizing hormone (LH), estradiol (E2), progesterone (PRG), prolactin (PRL) and total testosterone (TES), were normal. Negative for Chlamydia trachomatis (CT-DNA), ureaplasmaurealyticum (UU-DNA) and Neisseria gonorrhoeae (NG-DNA). The blood-routine exam was normal. Positive for HBsAb. Negative for hepatitis C, human immunodeficiency virus (HIV) and Treponema pallidum antibody. The karyotypes of the proband's parents, younger brother and wife were normal. A telephone follow-up in 2020 revealed no pregnancy. The pedigree is shown in Figure 1B.

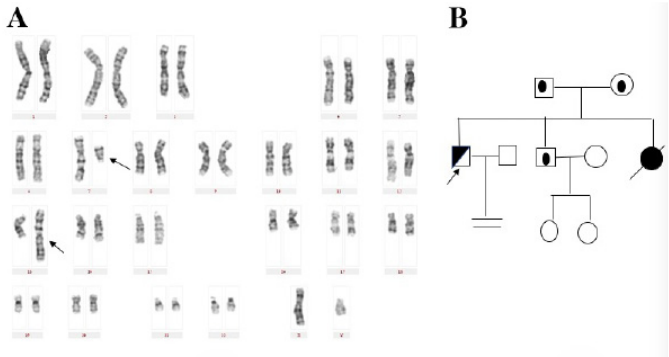

Fig.1: G-banding chromosome analysis and pedigree chart of case 1. **A.** Karyogram (Database No.3591) 46, XY, t(7;13) (p10;p10) dn and **B.** Pedigree chart.

Case 2: The proband was a 31-year-old man of Han nationality living in Qionghai City (Hainan Province, China). The patient was a company employee, his height was 170-cm. Normal phenotype and intelligence, three spontaneous

abortions within three years of marriage. In January 2014, he was admitted to the Second Affiliated Medical Reproductive Center of Hainan Medical University. Karyotype(Database No.3809):46,XY,t(12;17)(p10;q10) (Fig.2A). The blood-routine exam was normal. AB blood. Positive for RhD blood and RhC blood, negative for RhE blood. HBsAb was positive in hepatitis B screening, while the rest were negative for hepatitis C, HIV and Treponema pallidum antibody; negative for CT-DNA, UU-DNA and NG-DNA. Anti-sperm antibody was negative, and thalassemia genotyping was normal. The proband's wife had normal karyotype and became pregnant naturally as a telephone follow-up in 2014 showed. His older sister received a karyotype test, 46,XY,t(12;17)(p10;q10) at another hospital and had three spontaneous abortions. The pedigree is shown in Figure 2B.

Case 3: The proband was a 29-year-old female, of Han nationality living in Haikou City (Hainan Province, China). The patient was a company employee, her height was 160-cm. Normal in phenotype and intelligence, a year of marriage without pregnancy. The fallopian tubes were blocked and the uterus was 1.57 cm. In 2015, preimplantation genetic diagnosis (PGD) failed in the First Affiliated Medical Reproductive Center of Hainan Medical University. In January 2016, she was admitted to the Second Affiliated Medical Reproduction Center of Hainan Medical University. Karyotype (DatabaseNo.4914): 46,XX,t(2;6)(p10;q10)mat,21ps+ pat (Fig.3A). Cytology of the cervix was negative . All the biochemical indicators were normal. CT-DNA and UU-DNA were negative. The six sex hormones, including FSH, LH, E2, PRG, PRL and TES, were normal. The three items of hyperthyroidism were normal, namely, free triiodothyronine (FT3), free thyroid hormone (FT4) and the third-generation TSH. Insulin (INS) (reference range:2.8-24.7uU/mL):6.40uU/mL fasting, 105.50uU/mL one hour postprandial and 67.80uU/mL two hours postprandial. Blood glucose was normal (fasting, one hour postprandial and two hours postprandial). HBsAb was positive in hepatitis B screening, while the rest was negative. Negative for hepatitis C, HIV and Treponema pallidum antibody. Negative for the five items of eugenics. The anti-cardiolipin antibody was negative, and -A3.7 gene heterozygosity was detected in thalassemia genotyping. No pregnancy was found during a telephone follow-up in 2019. The husband's karyotype was normal, while the karyotypes of her mother and father were 46,XX,t(2;6)(p10;q10) and 46,XY,21ps+. The pedigree is shown in Figure 3B.

Comparison among WAT cases in 10 areas in China in pregnancy and childbirth history

Among the 29 probands from 10 areas in China, there were G85P9, wherein spontaneous abortions in the first trimester accounted for 48% (41/85); embryo arrest 31% (26/85); teratopia 6% (5/85); deaths 5% (4/85); amenorrhea 1% (1/85). There were two abortions, one case of unintended pregnancy and one case of biochemical one (Table 2).

Comparison of pregnancy history of whole arm translocation cases abroad

There were G25P5 probands from 7 families abroad, among which spontaneous abortion accounted for 72% (18/25) in early pregnancy, teratotypes accounted for 8% (2/25) and asthenospermia in 2 cases (Table 3).

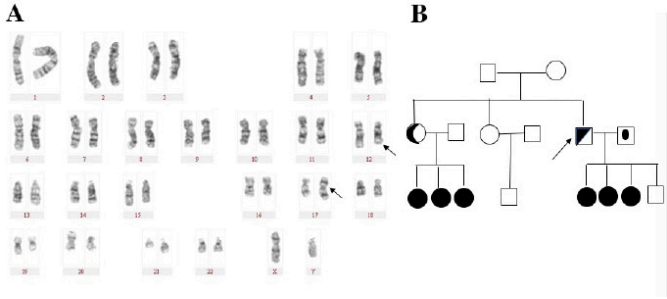

Fig.2: G-banding chromosome analysis and pedigree chart of case 2. **A.** Karyogram(DatabaseNo.3809) 46,XY,t(12;17)(p10;q10) and **B.** Pedigree chart.

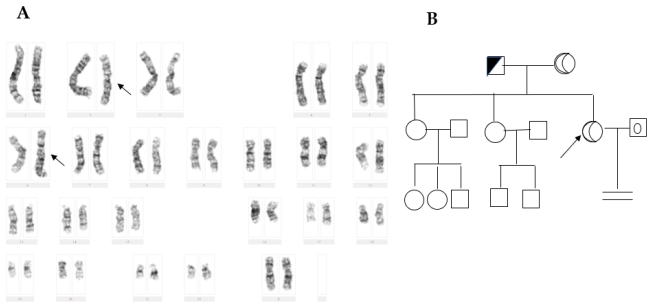

Fig.3: G-banding chromosome analysis and pedigree chart of case 3. **A.** Karyogram (DatabaseNo.4914) 46,XX,t(2;6)(p10;q10) mat,21ps+pat and **B.** Pedigree chart.

Table 2: Comparison among WAT cases in 10 areas in China in pregnancy and childbirth history

Literature	Chromosome karyotype	Area	Pregnancy history
Wang (1)	46,XX,t(1;6)(1pter→1p1-6q10→6qter;6pter→6p10-1q10→1qter)	Weihai City, Shandong Province	G3P1:Two spontaneous abortions and one deformed infant
Xiao (5)	46,XX,t(8;21)(p10;q10)(8pter→8p10::21q10→21qter;21pter→21p10::8q10→ 8qter)	Shenzhen City, Guangdong Province	G10P2:One postnatal death and eight spontaneous abortions
Lian (6)	46,XX,t(7;11)(7Petr→cen→11Pter;7qter→cen→11qter)	Shandong and Anhui Provinces	G2P0:Two spontaneous abortions
Liu (7)	46,XY,t(11;15)(11pter→cen→15pter;11qter→cen→15qter)	Shenzhen City, Guangdong Province	G2P0:Embryo arrest twice
Liu (7)	46,XY,t(1;16)(1pter→cen→16qter;1qter→cen→16pter)	Shenzhen City, Guangdong Province	G1P0:Unintended pregnancy
Tang (8)	46,XX,t(7,9)(7p9p.7q9q)(7pter→cen→9pter;7qter→cen→9qter)	Fuling Area, Sichuan Province	G3P0: Three spontaneous abortions
Zhang (9)	46,XY,t(11;15)(11pter→11p10::15p10→15pter;11qter→11q10::15q10→15qter)	Shenzhen City, Guangdong Province	G2P0:Two spontaneous abortions
Li (10)	46,XY,t(18;22)(p10;q10) mat	Linyi City, Shandong Province	G4P0: Four spontaneous abortions
Li (11)	46,XX,t(7:9)(p10;q10)	Beijing	Amenorrhea
Zhang (12)	46,XY,t(1;19)(1pter→1p11::19q11→19qter;19pter→19p11::1q11→1qter)	Guangzhou City, Guangdong Province	G2P1: One abortion
Mou (13)	46,XX.1(3;16)(3pter→3p10::16p10→16pter;3qter→3q10::16q10→16qter)	Nanchang City, Jiangxi Province	G0P0
Hu (14)	46,XY,t(7;12)(7pter→7p10::12p10→12pter::7qter→7q10::12q10→12qter)	Xuzhou City, Jiangsu Province	G1P0: Embryo arrest once
Liao (15)	46,XY,t(13;20)(13pter→13p10::20p10→20pter13qter→13q10::20ql0→20qter	Bengbu City, Anhui Province	G3P0: Three spontaneous abortions
Li (16)	46,XX,t(7:21)(7pter→7p10::21q10→21qter;21pter→21p10::7q10→7qter) mat,	Linyi City, Shandong Province	G2P0: Pregnant 1: hydrocephalus; Pregnant 2: fetal death
Li (16)	46,XX,t(7;14) (7pter→7p10::14p10→14pter::14qter→14q10::7q10→7qter) mat	Linyi City, Shandong Province	G4P0: Pregnancy 1: Abortion; Pregnancy 2: Embryo arrest; Pregnancy 3: Fetal death; Pregnancy 4: Spontaneous abortion
Li (16)	46,XY,t(9;18)(9pter→9pl0::18p10-18pter;18qter-18q10::9q10-9qter) mat	Linyi City, Shandong Province	G2P0: Pregnancy 1: Spontaneous abortion; Pregnancy 2: Omphalocele
Zhang (17)	46.XY,t(7;16)(7pter→7pl0::16q10→16qter;16pter→16p10::7q10→7qter) dn	Linyi City, Shandong Province	G3P0: Embryo arrest three times
Zhang (17)	46,XX,t(8;8)(8pter→8p10;:8p10→8pter;8qter-8q10::8q10-8qtcr)	Linyi City, Shandong Province	G5P0: Embryo arrest five times
Li (18)	46,XX,t(3;6)(3pter→3p10::6q10→6qter;6pter->6p10::3q10→3qter)pat	Linyi City, Shandong Province	G2P0: Two spontaneous abortions
Li (18)	46,XY,t(6;22)(6pter→6pl0::22q10→22qter;22pter→22pl0; :6q10→6qter)pat	Linyi City, Shandong Province	G2P0: Embryo arrest twice
Li (19)	46,XY,t(7;19)(7pter→7p10::19pl0→19pter;7qter→7q10::19q10→19qter)inv(9)(pter→pl2::q13→p12::q13→qter) mat	Linyi City, Shandong Province	G2P0: Two spontaneous abortions
Li (19)	46,XX,t(11;12(11pter→11p10::12pl0→12pter;11qter→11q10::12q10→12qter	Linyi City, Shandong Province	G2P0: Embryo arrest twice
Li (20)	46,XY,t(16;19)(16pter→16p10::19q10→19qter;19pter→19p10;:16q10→16qter)pat	Linyi City, Shandong Province	G2P0: Embryo arrest twice
Li (20)	46,XX,t(2;2)(2pter→2p10::2p10→2pter;2qter→2q10::2q10→2qter) dn	Linyi City, Shandong Province	G3P0: Embryo arrest three times
Li (21)	46,XY,t(18;22)(18pter→18p10::22q10→22qter;22pter→22p10::18q10→18qter) mat	Linyi City, Shandong Province	G4P0: Four spontaneous abortions
Li (21)	46,XY,t(18;22)(18pter→18p10::22q10→22qter;22pter→22p10::18q10→18qter) mat	Linyi City, Shandong Province	G2P2: The first baby girl had cleft lip and palate (CLP) and died a month later, due to eating difficulties. The second pregnancy was full-term normal delivery of a baby girl. She is now two years old, with normal intelligence and polydactylism.
Li (21)	46,XY,t(7;19)(7pter→7p10::19q10→19qter:19pter→19p10::7q10→7qter)pat	Linyi City, Shandong Province	G4P1: One spontaneous abortion and embryo arrest twice
Li (21)	46,XX,t(11;13)(11pter→11p10::13q10→13qter;13pter→13p10::11q10→11qter)pat	Linyi City, Shandong Province	G3P1: Embryo arrest twice
Liu (22)	46,XX,t(4;10)(4pter→4p10::10p10-10pter,4qter→4q10::10q10→10qter)	Linyi City, Shandong Province	G10P1: Six spontaneous abortions, induced labor once with single umbilical artery (SUA) and heart malformation, embryo arrest once and biochemical pregnancy once

Table 3: Comparison of pregnancy history of whole arm translocation cases abroad

Literature	Chromosome karyotype	Areas abroad	Pregnancy history
Safavi et al. (23)	46,XY,(13;18)(q10;q10)	Tehran, Iran	G1P1, one deformed infant
Fryns et al. (24)	46,XX,t(6plOq;6qlOp)	Leuven, Belgium	G4P1, Threespontaneous abortions
Fryns et al. (24)	46,XX,t(6plOq;6qlOp)mat	Leuven, Belgium	G4P1, Three spontaneous abortions
Vialard et al. (25)	46,XY,t(1;21)(q11;p13)	American	No refined disease
Vialard et al. (25)	46,XY,t(1;22)(q11;p11)mat	American	No refined disease
Tümer et al. (26)	46,XY,der(18),t(18;2)(p10;q10)pat	American	G2P1,one deformed infant
Smith et al. (27)	46, XX,t(1p5q;1q5p)	American	G9P2,7 spontaneous abortions
Smith et al. (27)	46,XX,-14,+derl4,t(10;14) (l0pl4q;14pl0q)mat	American	G5P01,5 spontaneous abortions

Discussion

The whole arm translocation is usually only a sporadic case report. However, there is no relevant literature on comparative statistics of a pregnancy and childbirth history with WAT. Chen et al. (28) reported and analyzed that the total detection rate of chromosomal abnormalities in patients with adverse pregnancies and childbirth histories in 20 provinces and cities in China and 16 countries was 5.62% (3,842/68,267), among which the incidence of autosomal balanced translocation was only 1.97% (1,325/68,267). Chromosome translocation is a significant cause of habitual abortion. Zhu (29) studied 42 balanced translocation carriers with a total of 90 pregnancies, among which 75 spontaneous abortions occurred during the first trimester, representing 83.4%. Chen (30) reported that the incidence of WATs was only 0.09% (3/3,353), and results of yunchun's study suggested that balanced chromosomal translocation carriers are associated with reproductive risks and a very high probability of abnormal pregnancy.

In this study, WATs was reported in 10 areas in China that were analyzed and their pregnancy and birth histories compared (1, 5-22). The 29 family probands had a total of 85 pregnancies and nine births. Specifically, spontaneous abortions accounted for 48% (41/85), while embryo arrest 31% (26/85); deformed infants 6% (5/85); deaths 5% (4/85). There were G25P5 (23-27), foreign family consents, among which spontaneous abortion in early pregnancy accounted for 72% (18/25), teratoma accounted for 8% (2/25), and azoospermia in 2 cases. WATs are relatively rare cytogenetic aberrations, mostly, these translocations are unbalanced accompanied by genomic imbalances (31). It has been reported (23) a rare cytogenetic variant of Monosomy 18p Syndrome as a consequence of WATs between chromosomes 13 and 18. Additionally, it has been reported (32) that, among 101 WAT cases, organized WATs seemed to occur more frequently than other translocations, which may explain that DNA promotes mismatches and unreasonably non homologous recombination. The three cases reported in this study also had an adverse pregnancy history for years, and case 1 even had azoospermia, indicating that WAT led to a severer pregnancy history.

As of December 2005, the Mitelman database (33, 34) included 933 cases of WAT chromosomes. WAT t(17;18) was detected in two cases (35) of acute monocytic leukemia and the conversion from acute to chronic granulocytic leukemia (CGL). Since these patients had either myeloid leukemia or myelodysplastic syndrome (MDS), it is suggested that t(17;18)(p10;q10) translocation is a novel nonrandom abnormality associated with myeloid cell proliferation. MDS(5;19)(p10;q10) cases (36). Moreover, der(5;19)(p10;q10) is a rare chromosomal abnormality of MDS that is genetically similar to 5q [del(5q)] deletion. These results indicate that der(5;19)(p10;q10) may play a significant role in the pathogenesis of high-risk MDS, because it is a rare but recurring translocation (36-38). One case (16) of MDS (refractory anemia) with karyotype 46,XY,+1,der(1;10)(q10;P10) resulted in abnormalities of trisomy 1q and monomer 10q, which may be necessary for tumor transformation. Clarifying the clinical features of myeloid neoplasms in patients with WATs would facilitate the elucidation of their tumorigenic mechanisms. But the pathogenesis of this cytogenetic anomaly still remains unresolved, an accumulation of cases with centromeric translocation may be still necessary.

Conclusion

When the parents are carriers of WAT, the family should be considered to have a high reproductive risk. Particularly, when the translocation area is a small chromosomal segment, the possibility of nonrandom involvement of some chromosomes in WAT and the risk of producing offspring with chromosomal abnormalities will increase. And this study is beneficial for better natal and prenatal care and better upbringing, conducting targeted inspections and guidance. The three cases of abnormal karyotype in this study have not yet been found in literature at home and abroad. This first report on the detection of the karyotype of human chromosomal aberration enriches clinical data on genetics for genetic counseling and prenatal diagnosis that serves as an important basis for research of the occurrence.

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Authors' Contributions

C.Y., L.Y., L.C.; Conceptualization, Methodology, Software, Writing, Reviewing and Editing, Writing the original draft, and Supervision. Z.Z., H.X., K.C.; Visualization and Investigation. All authors read and approved the final manuscript.

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