

Original Article

Factors influencing pregnancy termination decisions and a predictive model in women with chromosomal abnormalities detected through prenatal diagnosis

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Received May 14, 2026; Accepted July 31, 2026; Epub September 15, 2026; Published September 30, 2026

Abstract: Objective: To analyze the factors influencing pregnancy termination decisions in women with prenatally diagnosed chromosomal abnormalities and to develop a predictive model for termination risk. Methods: Retrospective clinical data were collected from 486 singleton pregnant women who received invasive prenatal diagnosis at West China Second Hospital between January 2023 and December 2024. Participants were divided into a termination group ($n=301$) and a continuation group ($n=185$) based on their final pregnancy management decision. Baseline data, prenatal diagnosis data and chromosome abnormality types of pregnant women were collected. Univariate and multivariate logistic regression analyses were used to identify factors influencing the decision to terminate pregnancy. A predictive model was developed based on these factors and internally validated using the Bootstrap method. Model discrimination, calibration, and clinical utility were assessed using receiver operating characteristic (ROC) curves, calibration curves, and decision-making curves (DCA). Results: Multivariate logistic regression showed that primiparity, autosomal aneuploidy, and sex chromosome mosaicism were independent risk factors for pregnancy termination, whereas chromosomal structural abnormalities were independent protective factors. The nomogram model demonstrated moderate discrimination and calibration. DCA showed that the net benefit was superior to the 'treat all' and 'treat none' strategies within the threshold probability range of 0.2-0.7; however, the benefit was limited, and external verification is required. Conclusion: Primiparity, autosomal aneuploidy and sex chromosome chimera were independent risk factors for termination of pregnancy, while chromosomal structural abnormalities were independent protective factors for termination of pregnancy. The nomogram showed acceptable discrimination and calibration, but its clinical net benefit was limited and the applicable risk range was narrow.

Keywords: Prenatal diagnosis, chromosomal abnormalities, terminate pregnancy, risk factors, predictive models

Introduction

Chromosomal abnormalities are a major cause of adverse pregnancy outcomes and birth defects [1, 2]. With the widespread adoption and increasing accuracy of prenatal diagnostic techniques, the detection rate of chromosomal abnormalities has risen significantly [3, 4]. However, the types of chromosomal abnormalities detected prenatally are diverse, including autosomal aneuploidy, sex chromosome numerical abnormalities, structural chromosomal abnormalities and mosaicism. The impact of these abnormalities on fetal growth and development vary considerably [5, 6]. When confronted with a confirmed diagnosis, pregnant

women and their families must make a termination decision within a limited time frame and with often incomplete information. This decision is not only affected by medical prognosis, but also involves multiple factors such as psychology, ethics, family and society. It is one of the most challenging links in prenatal genetic counseling.

Existing research, both domestically and internationally, on pregnancy termination decisions in women with chromosomal abnormalities has largely been limited to describing outcomes for specific abnormality types [7] or follow-up analyses of particular prenatal diagnosis indication [8]; and there has been no systematic compari-

son of the relative influence of different chromosomal abnormality types on the decision-making process. Moreover, the existing research has not yet formed a unified risk assessment system, which has difficulty meeting the needs of individualized consultation. A predictive model can quantify and integrate independent risk factors identified by multivariate analysis and visually present the predicted probability of pregnancy termination. In recent years, predictive modeling has shown good application value in obstetric risk prediction [9, 10]. Nevertheless, no nomogram has yet been developed to quantify the likelihood of pregnancy termination in women carrying fetuses with chromosomal abnormalities.

Based on this, the present study aimed to retrospectively analyze the clinical data of pregnant women with chromosomal abnormalities in prenatal diagnosis, analyze the influence of different types of chromosomal abnormalities and other factors on the decision to terminate pregnancy, identify independent influencing factors, and construct a nomogram prediction model to provide a quantifiable decision-making aid for prenatal genetic counseling.

Materials and methods

Research subjects

The clinical data of 486 pregnant women with prenatal diagnosis of chromosomal abnormalities admitted to West China Second Hospital from January 2023 to December 2024 were retrospectively collected. Inclusion criteria: (1) Underwent prenatal diagnosis (amniocentesis) during the second trimester (19-22+6 weeks) and had a confirmed chromosomal abnormality [11, 12]; (2) Singleton pregnancy; (3) Maternal age ≥ 20 years; (4) A definitive pregnancy outcome; (5) Complete clinical records, prenatal diagnostic reports and follow-up data on pregnancy outcome. Exclusion criteria: (1) Multiple pregnancies; (2) Concurrent severe cardiac, hepatic, renal or hematological disorders, or active autoimmune disease; (3) Concurrent severe mental illness; (4) Complications including severe pre-eclampsia, severe gestational diabetes, acute infections during pregnancy, or severe hemorrhage; (5) Fetuses with severe structural malformations (e.g. anencephaly, severe spina bifida, major cardiac or vascular malformations) or clear evidence of non-chro-

mosomal inherited metabolic disorders in addition to chromosomal abnormalities; (6) Cases with missing key clinical indicators or an unclear pregnancy outcome. This study was approved by the ethics committee of West China Second Hospital.

Grouping method

All pregnant women were divided into two groups based on their final decision regarding the pregnancy:

Termination group ($n=301$): This comprised all cases in which, following prenatal diagnosis of fetal chromosomal abnormalities and thorough genetic counselling, the woman actively chose to terminate the pregnancy by induced labor. Continuation group ($n=185$): Cases in which the family refused to terminate the pregnancy and chose to continue the pregnancy until delivery [13-18]. This study followed participants only until one month postpartum and did not evaluate long-term neurodevelopmental or reproductive outcomes.

Data collection

This study was a retrospective analysis. The clinical data of all subjects were uniformly extracted and collated through the hospital electronic medical record system, and the prenatal diagnosis center follow-up registration system. Two professionally trained researchers independently completed data collection.

(1) Baseline data: Collect demographic characteristics of pregnant women and basic information on their pregnancies, including age, parity, estimated gestational age, and medication history.

(2) Prenatal diagnosis-related data: Indications for prenatal diagnosis (advanced maternal age, high-risk first-trimester screening, high-risk non-invasive genetic testing, adverse obstetric history, or maternal request) and origin of abnormalities (de novo mutation, paternally inherited, maternally inherited, or unknown).

(3) Types of chromosomal abnormalities: Based on karyotype analysis reports, fetal chromosomal abnormalities were classified into four major categories: structural chromosomal abnormalities, autosomal aneuploidy, sex chro-

Prenatal chromosome abnormality terminate pregnancy prediction

Table 1. Comparison of baseline data between the two groups

Indicator	Termination group (n=301)	Continuation group (n=185)	χ^2/t	P
Age, year	30.91±4.74	29.91±5.04	2.204	0.028
Parity			2.587	0.108
Primiparous	169 (56.15)	90 (48.65)		
Multiparous	132 (43.85)	95 (51.35)		
Gestational age, week	15.65±2.37	16.30±2.80	2.737	0.006
Medication history			0.038	0.847
Yes	92 (30.56)	55 (29.73)		
No	209 (69.44)	130 (70.27)		

between the two groups ($P < 0.05$) (**Table 1**).

Comparison of prenatal diagnosis-related data between the two groups of pregnant women

Comparisons of the indications for prenatal diagnosis and the sources of chromosomal abnormalities between the two groups of pregnant women revealed statistically

mosome numerical abnormalities, and sex chromosome mosaicism [19, 20].

Statistical analysis

All data analyses were performed using SPSS 27.0 (IBM Corp., Armonk, New York, USA). All continuous variables were normally distributed, and were expressed as mean $\pm$ standard deviation (SD); between-group comparisons were made using the independent sample t-test. Count data were expressed as frequency and percentage [n (%)], and chi-square test was used for comparison between groups. Univariate logistic regression was used to evaluate the association between chromosomal abnormality types and pregnancy termination. Multivariate logistic regression was then applied to identify independent factors influencing the termination decision. A Nomogram prediction model was constructed based on the identified factors, and the Bootstrap method ($B=1000$) was used for internal verification. The discrimination of the model was evaluated by the receiver operating characteristic (ROC) curve. Model calibration was assessed using the calibration curve and the Hosmer-Lemeshow test, and clinical utility was evaluated using decision curve analysis (DCA). $P < 0.05$ was considered statistically significant.

Results

Comparison of baseline data between the two groups

There was no statistically significant difference in parity and medication history between the two groups of pregnant women ($P > 0.05$). There were statistically significant differences in maternal age and diagnostic gestational age

significant differences ($P < 0.05$). In the termination group, advanced maternal age was the most common indication for prenatal diagnosis and was significantly more frequent than in the continuation group. In the continuation group, the proportions of cases attributed to a history of adverse pregnancy or childbirth, and the mother's active request for prenatal diagnosis were higher than in the termination group. In terms of causes of abnormalities, the proportion of new mutations in the termination group was significantly higher than in the continuation group, whereas the proportions of paternally and maternally inherited abnormalities were lower than those in the continuation group (**Table 2**).

Distribution of chromosomal abnormalities in pregnant women

This study categorized fetal chromosomal abnormalities into four types: structural chromosomal abnormalities, autosomal aneuploidy, sex chromosome numerical abnormalities, and sex chromosome mosaicism. Of these, structural chromosomal abnormalities accounted for 102 cases, and were the most common type of abnormality in the continuation group. Autosomal aneuploidy accounted for 176 cases, representing the most common type of abnormality in the termination group (**Table 3**).

Multivariate logistic regression analysis of factors influencing the decision to terminate pregnancy in women with chromosomal abnormalities

With pregnancy decision as the dependent variable, variables with $P < 0.05$ in the univariate analysis, along with other potential confounding factors, were included in a multivariate

Prenatal chromosome abnormality terminate pregnancy prediction

Table 2. Comparison of prenatal diagnosis relevant information between the two groups

Indicator	Termination group (n=301)	Continuation group (n=185)	χ^2	P
Diagnostic criteria			18.443	0.001
Pregnant woman's request	7 (2.33)	9 (4.86)		
Advanced maternal age	125 (41.53)	65 (35.14)		
High-risk from DSS	70 (23.26)	44 (23.78)		
High-risk from NIPT	78 (25.91)	34 (18.38)		
Adverse pregnancy and childbirth history	21 (6.98)	33 (17.84)		
Origin of abnormalities			9.322	0.025
Unknown	14 (4.65)	11 (5.95)		
New mutation	237 (78.74)	123 (66.49)		
Inherited from father	27 (8.97)	28 (15.14)		
Inherited from mother	23 (7.64)	23 (12.43)		

Note: DSS, down syndrome screening; NIPT, non-invasive prenatal testing.

Table 3. Distribution of chromosomal abnormalities

Type	Termination group (n=301)	Continuation group (n=185)	χ^2	P
Structural chromosomal abnormalities	30 (9.97)	72 (38.92)	57.916	<0.001
Autosomal aneuploidy	136 (45.18)	40 (21.62)	27.535	<0.001
Sex chromosome aneuploidy	72 (23.92)	50 (27.03)	0.588	0.443
Sex chromosome mosaicism	63 (20.93)	23 (12.43)	5.681	0.017

logistic regression analysis. The results showed that primiparity ($OR=1.764$, 95% CI : 1.093-2.847), autosomal aneuploidy ($OR=2.125$, 95% CI : 1.170-3.861) and sex chromosome mosaicism ($OR=2.757$, 95% CI : 1.320-5.761) were independent risk factors for the decision to terminate the pregnancy, whilst structural chromosomal abnormalities were an independent protective factor against termination ($OR=0.296$, 95% CI : 0.151-0.583). After multivariate adjustment, variables such as maternal age, gestational age at diagnosis, history of medication during pregnancy, various indications for prenatal diagnosis, source of chromosomal abnormalities and sex chromosome numerical abnormalities were not statistically significant (all $P>0.05$) (**Table 4**).

Development of a predictive model

Based on the risk factors identified through multivariate logistic regression analysis (Parity, chromosomal structural abnormalities, autosomal aneuploidy and sex chromosome mosaicism), a nomogram model was developed to predict the risk of pregnancy termination in pregnant women with chromosomal abnormalities (**Figure 1**).

ROC curves for factors influencing the decision to terminate pregnancy in women with chromosomal abnormalities

The predictive model was internally validated using the Bootstrap method ($B=1000$). Analysis of the ROC curve revealed that the model achieved an AUC of 0.725 (95% CI : 0.671-0.780) on the training set and an AUC of 0.656 (95% CI : 0.555-0.757) on the validation set (**Figure 2**).

Calibration of the predictive model and decision curve analysis

The calibration curve results show that the calibration curves for both the training set (**Figure 3A**) and the validation set (**Figure 3B**) were relatively close to the standard curve, suggesting that the model's predicted probability of pregnancy termination is in good agreement with the actual observed rate of pregnancy termination. The DCA curve (**Figure 4**) was used to assess the model's clinical net benefit at different probability thresholds. The results showed that the net benefit of the model was slightly better than that of the 'all termination' or 'all continuation of pregnancy' strategy only

Prenatal chromosome abnormality terminate pregnancy prediction

Table 4. Multivariate logistic regression analysis of factors influencing the decision to terminate pregnancy in women with chromosomal abnormalities

Variables	Univariate analysis			Multivariate analysis		
	β	P	OR (95% CI)	β	P	OR (95% CI)
Age	0.021	0.357	1.021 (0.976-1.068)			
Parity						
Multiparous			1.000 (Reference)			1.000 (Reference)
Primiparous	0.523	0.020	1.688 (1.086-2.623)	0.568	0.020	1.764 (1.093-2.847)
Gestational age	-0.072	0.094	0.930 (0.855-1.012)			
Medication history						
Yes	-0.095	0.694	0.909 (0.566-1.461)			
No			1.000 (Reference)			
Pregnant woman's request	-0.989	0.089	0.372 (0.119-1.163)			
Advanced maternal age	0.030	0.897	1.030 (0.655-1.620)			
High-risk from DSS	0.024	0.927	1.024 (0.614-1.709)			
High-risk from NIPT	0.472	0.082	1.603 (0.943-2.725)			
Adverse pregnancy and childbirth history	-0.601	0.093	0.548 (0.271-1.107)			
Unknown	-0.029	0.953	0.971 (0.367-2.572)			
New mutation	0.388	0.119	1.474 (0.905-2.400)			
Inherited from father	-0.288	0.419	0.750 (0.373-1.506)			
Inherited from mother	-0.480	0.170	0.619 (0.312-1.229)			
Chromosomal structural abnormalities	-1.749	<0.001	0.174 (0.097-0.311)	-1.217	<0.001	0.296 (0.151-0.583)
Autosomal aneuploidy	0.897	<0.001	2.452 (1.500-4.007)	0.754	0.013	2.125 (1.170-3.861)
Sex chromosome numerical abnormalities	-0.215	0.393	0.806 (0.492-1.322)			
Sex chromosome mosaicism	0.951	0.004	2.589 (1.367-4.906)	1.014	0.007	2.757 (1.320-5.761)

Note: OR, odds ratio; CI, confidence interval.

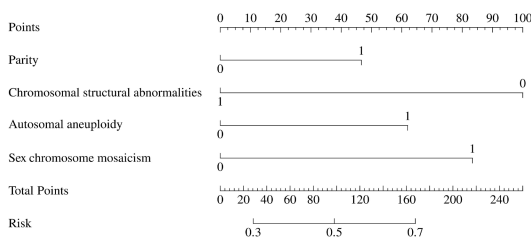


Figure 1. Nomogram for predicting pregnancy termination risk. Note: Parity (Primiparous = 1, Multiparous = 0); Chromosomal structural abnormalities, Autosomal aneuploidy and Sex chromosome mosaicism (Yes = 1, No = 0).

in the medium risk threshold range of 0.2-0.7. At the threshold probability of 0.4, the net benefit of the model is only about 0.1, and the benefit range is limited. When the risk threshold was <0.1 or >0.8, the model's net benefit essentially approached zero, making it difficult to provide effective incremental guidance for decision-making. On the whole, the application range of the model is relatively narrow, and it can only be used as a limited auxiliary reference tool. Clinical decision-making cannot rely solely on the results of this model.

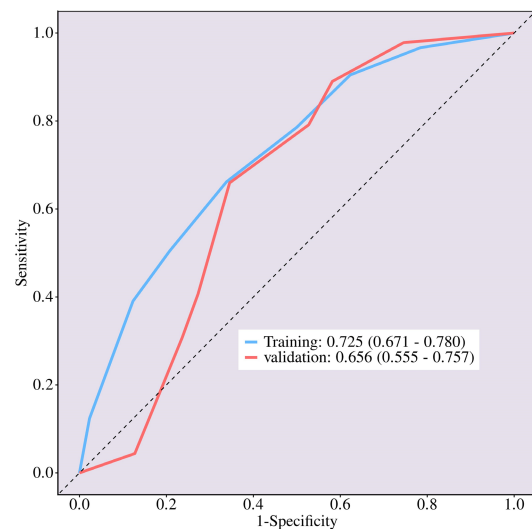


Figure 2. ROC curves for factors influencing pregnancy outcomes in women with chromosomal abnormalities.

Discussion

This study analyzed the clinical data of pregnant women with chromosomal abnormalities identified through antenatal diagnosis to inves-

Prenatal chromosome abnormality terminate pregnancy prediction

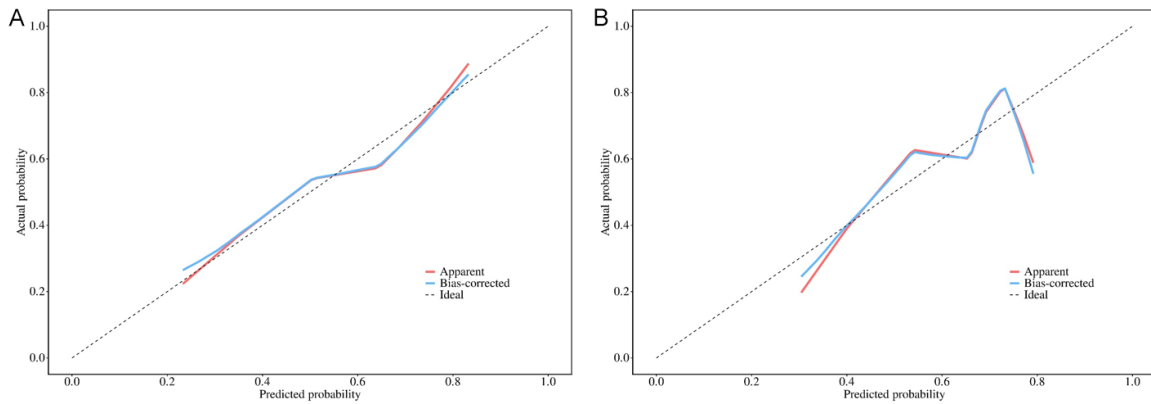


Figure 3. Calibration curves for the prediction model in the training set (A) and validation set (B).

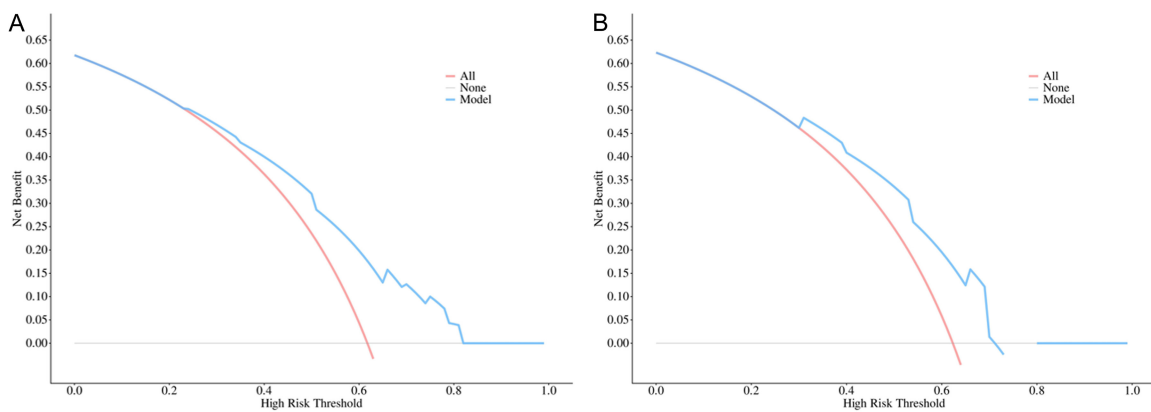


Figure 4. Decision analysis curve for the prediction model in the training set (A) and validation set (B).

tigate the independent factors influencing the decision to terminate pregnancy. Multivariate logistic regression analysis revealed that primiparity, autosomal aneuploidy and sex chromosome mosaicism were independent risk factors for the decision to terminate pregnancy, whilst structural chromosomal abnormalities were an independent protective factor against termination. The nomogram model constructed based on these variables showed moderate discriminatory power and calibration, but its clinical net benefit was limited; it may provide a quantitative decision-making reference for prenatal genetic counseling, though with notable caveats.

This study found that autosomal aneuploidy is one of the strongest risk factors contributing to the decision to terminate a pregnancy, with a termination rate as high as 77.27%. This finding is consistent with previous studies [21, 22]. Autosomal aneuploidies such as trisomy 21, trisomy 18 and trisomy 13 typically involve critical

chromosomal regions, resulting in severe intellectual disability, multisystem structural malformations, and poor postnatal survival. The clinical prognosis is generally poor, and the vast majority of pregnant women and their families tend to opt for termination of pregnancy after receiving full information [23, 24]. It is worth noting that sex chromosome mosaicism in this study had the most prominent effect on the decision-making of termination of pregnancy ($OR=2.757$). The phenotypic spectrum of mosaicism is broad due to variable distribution of abnormal cell lines across different tissues, ranging from near-normal to classic syndromic presentations [25]. This uncertainty often makes pregnant women and their families bear greater psychological pressure in decision-making, and they thus tend to choose to terminate the pregnancy. Moreover, the difficulty in accurately predicting prognosis for mosaic cases may bias decision-making toward termination. This finding suggests that sex chromosome mosaicism warrants greater attention in

genetic counseling, and the prognostic uncertainty should be communicated objectively to avoid decision bias caused by information ambiguity.

In contrast to the risk factors mentioned above, chromosomal structural abnormalities represent an independent protective factor in the decision to terminate a pregnancy, with a significantly higher rate of pregnancy continuation compared to other types of abnormalities. In this study, the pregnancy termination rate in the structural abnormality group was only 29.41%, significantly lower than that in the autosomal aneuploidy and sex chromosome chimera groups. This result may be related to the composition of structural abnormalities; certain structural abnormalities, such as balanced translocations, inversions, and small-segment deletions or duplications, may not affect normal fetal development if they do not disrupt key genes or cause dosage imbalance [26]. In addition, some structural abnormalities are inherited from one of the parents with normal phenotype, and their potential pathogenicity is further reduced. Therefore, in clinical practice, decisions regarding structural chromosomal abnormalities should integrate parental karyotype analysis, high-resolution ultrasound, and pathogenicity interpretation, so as to avoid unnecessary pregnancy termination.

With regard to obstetric factors, primiparity was the only statistically significant socio-demographic factor among pregnant women in this study. The risk of pregnancy termination decision was significantly higher among primiparous women than among multiparous women. This is because multiparous women, with prior childbirth experience, may have a better understanding of fetal conditions and the demands of parenting, and their families have greater capacity to cope. In contrast, families experiencing their first pregnancy often lack knowledge of genetic disorders and birth defects; they find it difficult to accept the possibility of congenital abnormalities in the fetus, have lower psychological resilience, and are more likely to opt for termination of pregnancy [27]. It is worth noting that baseline variables such as maternal age and gestational age at diagnosis were statistically significant in univariate analysis but not in multivariate analysis. Specifically, although there were significant differences in age and diagnostic gestational age between

groups in univariate analysis, the independent effect was no longer significant after adjusting for chromosome abnormality type as a confounding factor. The high rate of pregnancy termination in pregnant women with advanced age is essentially due to the close relationship between advanced age and the risk of aneuploidy, rather than the advanced age itself directly driving decision-making [28, 29]. Similarly, the proportion of termination of pregnancy is higher in patients with early diagnosis of gestational age, mainly because the abnormal types with strong pathogenicity are mostly screened and diagnosed in the early stage of pregnancy, and the independent predictive effect of gestational age is diluted after correcting the abnormal types. In terms of indications for prenatal diagnosis, advanced age and high NIPT risk were no longer significant after adjustment in the multivariate model. The reason is also that these indications are highly correlated with the types of chromosomal abnormalities. The high risk of NIPT itself is highly suggestive of aneuploidy [30, 31], the effect has been captured by the model through abnormal type variables.

Based on the above influencing factors, a prediction model was constructed in this study. The model integrated parity and three main types of chromosomal abnormalities. After Bootstrap internal verification, the AUC of the training set was 0.725, and the AUC of the validation set was 0.656. The calibration curve is close to the ideal standard curve, suggesting that the model has a certain degree of discrimination and calibration. The DCA showed that the model's net benefit exceeded that of the 'treat all' and 'treat none' strategies only within the intermediate risk threshold range of 0.2-0.7, and the benefit range is limited (the net benefit is about 0.1 at the threshold of 0.4), with no additional clinical benefit at very low or high risk thresholds. This indicates that the model's clinical utility is limited; it should not be used as a standalone decision-making tool but rather as supplementary reference in genetic counseling.

There are some limitations in this study: First, this was a single-center retrospective study, which may be subject to selection bias in case selection, diagnostic procedures, and genetic counseling practices. Second, the model included only maternal baseline characteristics, prenatal diagnosis indications, and chromo-

somal abnormality categories, and did not account for other important confounders such as fetal sex, ultrasound soft markers/structural abnormalities, mosaicism level, maternal psychological state, family financial situation, and partner's understanding. These factors may influence the family's decision, and their omission could affect the model's predictive accuracy. The follow-up time of this study was only 1 month after delivery, and the long-term prognosis of long-term neurodevelopment, physical growth and reproductive health of newborns who survived after continuing pregnancy was not tracked. Long-term prognostic information is an important reference for decision-making, and the long-term prediction efficiency of the model needs to be further verified. Future multi-center, large-scale prospective cohort studies including data from different regions and hospital levels are needed for external validation of the model. Refined subtyping of chromosomal abnormalities and integration of fetal ultrasound findings, maternal complications, and psychosocial factors could improve model performance. Extended follow-up is also warranted to evaluate long-term neurodevelopmental and physical outcomes.

Conclusion

Primiparity, autosomal aneuploidy and sex chromosome chimera were independent risk factors for termination of pregnancy, while chromosomal structural abnormalities were independent protective factors for termination of pregnancy. The discrimination and calibration of the constructed Nomogram model are acceptable, but the clinical net benefit is limited and the applicable risk interval is narrow, so it can only be used as an auxiliary reference tool for prenatal genetic counseling.

Disclosure of conflict of interest

None.

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