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Current Status and Future Role of Fetal Heart Disease Diagnosis

Before 22 Weeks’ Gestation at Our Institution.

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Introduction

In Japan, under the Maternal Health Protection Law, induced abortion is permitted before 22 weeks of pregnancy for specified reasons. Advances in echocardiographic equipment have enabled earlier diagnosis of fetal heart disease. Early prenatal diagnosis can influence the critical decision of pregnancy termination, requiring accurate and sensitive care. Our facility performs early fetal echocardiography to provide prenatal diagnosis and counseling for congenital heart disease (CHD). The relationship between early prenatal diagnosis of congenital heart disease, patient background, and decision-making regarding pregnancy termination remains fully unclarified.

Objective

Factors associated with the choice of pregnancy termination in cases with CHD diagnosed before 22 weeks of gestation were evaluated.

Methods

Subjects : Subjects who fulfilled the following three conditions:
• Ftal echocardiography performed at Kindai University Hospital between September 2016 and December 2025
• Geestational age < 22 weeks (at diagnosis)
• Diagnosed with congenital heart disease (CHD)

Primary Outcome: Occurrence of elective termination of pregnancy

Primary Analytical Factors: the following three factors
① Disease severity (3 groups based on specific diagnosis) (Table1)
② Gestational age at diagnosis
③ Maternal age

Supplementary / Sensitivity Analyses:
• Disease Severity: Evaluated using expected circulation (Fontan circulation vs. biventricular repair) as a supplementary index of disease severity
• Chromosomal Abnormalities: Evaluated only in supplementary analyses due to a high proportion of missing data, despite their clinical significance.

Results

A total of 104 CHD cases diagnosed prior to 22 weeks of gestation during the study period were identified. After excluding 8 cases with unknown pregnancy outcomes, 96 cases (32 terminations and 64 continued pregnancies) were included in the univariate analysis. Subsequently, a complete case analysis was performed for multivariate analysis on 93 cases, excluding 3 cases with unknown disease severity. (Table2)

◆ Univariate Analysis of Baseline Characteristics (n = 96) (Figure3)
• Gestational Age at Diagnosis: Continued: 18 weeks vs. Termination: 19 weeks
• Maternal Age: 35 vs. 33 years (Median) — *No apparent difference*
• Advanced Maternal Age (AMA): 53.1% (Continued) vs. 34.4% (Termination)
→ *Higher proportion of AMA in the continued pregnancy group*
• Key Associated Factors with Termination:
•Fontan circulation
•High-risk disease severity

◆ Chromosomal Evaluation (Supplementary Analysis)(n = 96) (Table4)
• High Missing Data Rate: 41.7% (n = 40) missing / unperformed
• Normal Karyotype (n = 49):
•Continued pregnancy: n = 35 (71.4%)
•Termination: n = 14 (28.6%)
→ *Trend toward pregnancy continuation when normal*
• Abnormal Karyotype in Termination Group: 9.4% (n = 3/32)

◆Multivariate Logistic Regression Analysis (n = 93) (Table5)
• Adjusted Model Variables: Disease Severity, Gestational Age at Diagnosis, Maternal Age
• Key Finding:
•High-Risk Disease Severity: Significantly associated with higher rates of pregnancy termination (indicated by adjusted OR and 95% CI).
•Gestational Age & Maternal Age: Not independently associated with termination after adjustment.

◆Supplementary Analysis: Multivariable Model (n = 93) (Table6)
• Variables: Fontan Circulation, Gestational Age at Diagnosis, Maternal Age
• Key Finding:
Fontan Circulation: Showed a trend toward higher rate of pregnancy termination based on the odds ratio, but was not statistically significant.

Discussion

Determinants of Pregnancy Termination
• Main Finding of This Study:
Disease Severity was suggested to be the primary factor determining pregnancy termination.
• Comparison with General Prenatal Diagnosis & Literature:
• General Prenatal Diagnosis: Fetal "chromosomal abnormalities" are widely known as the main driver for termination.
→ In our study, however, chromosomal abnormalities had a **weak impact** on the decision.
• Domestic Reports (Japan): "Critical CHD" and "co-existing extracardiac/genetic anomalies" are independently associated with termination.
Nakao M, Ikeda T , et al ; Sci Rep 2024;14:15055
• Clinical Insight:
• In prenatally diagnosed CHD (< 22 weeks), the severe prognosis of the cardiac disease itself—rather than chromosomal status alone—serves as the ultimate determinant in parental decision-making.
→There is a possibility that patients had already chosen termination during first-trimester screening (e.g., NIPT), as well as a risk of underestimation due to 41.7% missing data.

Need for mindful counseling:
• Healthcare providers must deliver prenatal counseling recognizing that disease severity may drive the decision for pregnancy termination.
Clinical guidelines emphasize the importance of informed consent both before and after genetic testing, as well as family care following non-directive disclosure, practices that are already widely implemented by many healthcare providers.

Establishment of Support Systems:
• Need to build a framework that enables families to make optimal decisions during the brief timeframe (a few weeks) between diagnosis and termination.
• Shift toward Shared Decision Making (SDM) & Multidisciplinary Care
• Need for SDM over Traditional IC:
• Informed Consent (IC): A unidirectional process where physicians explain medical information and patients grant consent.
• Shared Decision Making (SDM): An interactive process where medical and social information is shared, deliberating and deciding treatment plans together through dialogue.
• Multidisciplinary Collaboration: Essential to support families in making optimal decisions during the limited timeframe.

Limitation

• Single-center, retrospective design with a small sample size.
• High rate of missing data regarding the presence of chromosomal abnormalities.
• Key confounding factors (legal limit of gestational age, history of infertility treatment, family socioeconomic background, and institutional counseling policies) were unmeasured.
• Non-significant findings reflect inconclusive results rather than an absence of association.

Conclusion

● In fetal cardiac disease diagnosed before 22 weeks of gestation, disease severity was the primary factor determining the choice of pregnancy termination.

● Decision-making support focused on disease severity is desirable.

● To this end, a Shared Decision Making (SDM)-oriented process, which has gained attention in recent years, may be useful.

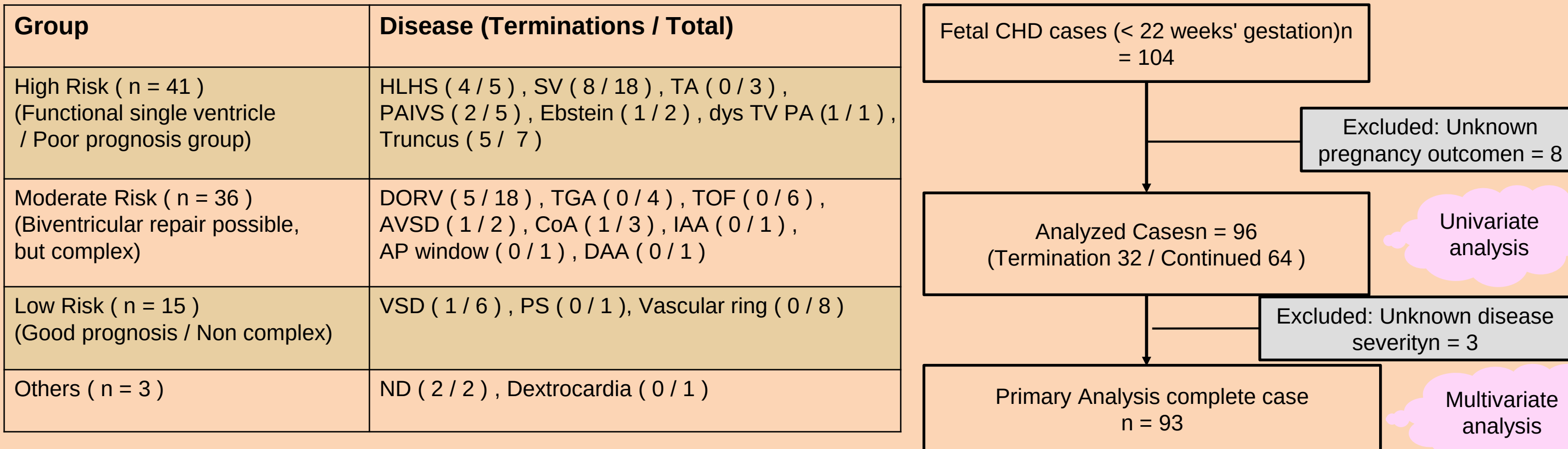


Table1. 3 groups based on specific diagnosis
Classified into 3 groups based on functional status and prognosis due to the absence of a standardized severity scale. Diagnoses are listed above.

Table2. Selection of Study Population and Analytical Methods

Factor	Total	Continued	Termination	p
n	96	64	32	
Gestational age at diagnosis(weeks)	19 [18.0 – 20.0]	18.0 [18.0 – 20.0]	19.0 [17.8 – 19.0]	0.277
Maternal age (years)	34.0 [31.0 – 37.0]	35.0 [30.8 – 38.2]	33.0 [31.0 – 36.0]	0.288
Advanced maternal age (≥ 35 years)	45 (46.9 %)	34 (53.1 %)	11 (34.4 %)	0.129
Fontan circulation	34 (37.8)	18 (29.5)	16 (55.2)	0.034
Disease severity				0.002
Low risk	16 (17.2)	14 (22.2)	2 (6.7)	
Moderate risk	36 (38.7)	29 (46.0)	7 (23.3)	
High risk	41 (44.1)	20 (31.7)	21 (70.0)	

Values are median [IQR] or n (%). Continuous variables were analyzed using the Wilcoxon test, and categorical variables using Fisher's exact test.

Table3. Results: Patient Characteristics and Univariate Analysis (Termination vs. Continued Pregnancy)

Factor	全体	Continued	Termination	p
n	96	64	32	
Chromosomal test				0.577
normal	49 (51.0)	35 (54.7)	14 (43.8)	
abnormal	7 (7.3)	4 (6.2)	3 (9.4)	
ND	40 (41.7)	25 (39.1)	15 (46.9)	
Extracardiac anomalies	10 (11.6)	3 (5.2)	7 (25.0)	0.020

Values are median [IQR] or n (%). Continuous variables were analyzed using the Wilcoxon test, and categorical variables using Fisher's exact test.

Table4. Results: Patient Characteristics(Chromosomal test &Extracardiac anomalies) and Univariate Analysis (Termination vs. Continued Pregnancy)

Factor	aOR	95 % CI	p
Moderate risk (VS Low risk)	1.55	0.31 – 11.68	0.622
High risk (VS Low risk)	6.87	1.59 – 48.37	0.020
Gestational age at diagnosis(weeks)	0.90	0.63 – 1.27	0.54
Maternal age (years)	1.01	0.90 – 1.14	0.836

(n = 93)

Table5. Primary Analysis complete case
Multivariate Logistic Regression Analysis for Pregnancy Termination.

Factor	aOR	95 % CI	p
Fontan circulation (vs. Biventricular repair)	2.43	0.88 – 6.94	0.090
Gestational age at diagnosis(weeks)	0.87	0.61 – 1.22	0.417
Maternal age (years)	0.98	0.88 – 1.09	0.702

Table6. Supplementary Analysis: Multivariable Model including Fontan Circulation
(n = 93)