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MedEst|2026 | Vol.6: e523| ISSN: 2789-7567 |RNPS: 2524| BRIEF COMMUNICATION

Risk factors and repercussions on family dynamics of prenatal diagnosis of congenital heart disease

Factores de riesgo y repercusiones en la dinámica familiar del diagnóstico prenatal de cardiopatías congénitas

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RESUMEN

Introducción: las cardiopatías congénitas son la anomalía congénita más frecuente, con prevalencia global de 8 a 12 por 1.000 nacidos vivos. Este estudio evaluó factores de riesgo y repercusiones familiares del diagnóstico prenatal de cardiopatías congénitas en áreas de los policlínicos Aleida Fernández, Cristóbal Labra y Elpidio Berovides, desde la Atención Primaria de Salud. **Objetivo:** identificar los factores de riesgo asociados al diagnóstico prenatal de cardiopatías congénitas en mujeres embarazadas atendidas en tres policlínicos del municipio La Lisa, Cuba, durante 2024. **Métodos:** estudio descriptivo transversal (2024) en 18 consultorios de tres policlínicos (n=360 embarazadas; muestreo estratificado). Se emplearon encuesta adaptada de WHO, Escala FAD (α=0,89) y ecografía Doppler (sensibilidad 87%).Análisis descriptivo, correlación Pearson (r) y OR (SPSS v.26, $p < 0,05$). **Resultados:** prevalencia de cardiopatías congénitas: 9,2% (n=33; septales 55%). Riesgos: edad > 35 años (OR 3,1; IC 95% 1,8-5,4), tabaquismo (r=0,45; $p < 0,01$). Dinámica familiar: FAD media 2,6 ± 0,9 en casos versus 1,9 ± 0,7 en controles (*diferencia de medias: 0,7; $p < 0,001$*); estrés económico 70% (OR 3,5). La detección temprana en APS alcanzó 65% y redujo ansiedad en 28%. **Conclusiones:** las cardiopatías congénitas prenatales correlacionaron con riesgos modificables, alterando la cohesión familiar. La Atención Primaria de Salud facilitó detección y apoyo, mitigando impactos en 30%; se recomiendan programas integrales.

ABSTRACT

Introduction: congenital heart defects are the most common congenital anomaly, with a global prevalence of 8 to 12 per 1,000 live births. This study evaluated risk factors and family repercussions associated with the prenatal diagnosis of congenital heart defects in the catchment areas of the Aleida Fernandez, Cristobal Labra, and Elpidio Berovides polyclinics, from the perspective of Primary Health Care. **Objective:** to identify the risk factors associated with the prenatal diagnosis of congenital heart disease in pregnant women treated in three polyclinics in the municipality of La Lisa, Cuba, during 2024. **Methods:** a descriptive cross-sectional study (2024) was conducted across 18 medical offices within three polyclinics (n = 360 pregnant women; stratified sampling). The instruments employed included a WHO-adapted survey, the FAD Scale (α = 0,89), and Doppler ultrasound (87% sensitivity). Data analysis involved descriptive statistics, Pearson correlation (r), and Odds Ratios (OR) (SPSS v.26; $p < 0,05$). **Results:** the prevalence of congenital heart defects was 9.2% (n = 33; 55% of which were septal defects). Identified risk factors included maternal age >35 years (OR 3,1; 95% CI: 1,8–5,4) and smoking (r = 0,45; $p < 0,01$). Regarding family dynamics, the mean FAD score was 2,6 ± 0,9 in cases versus 1,9 ± 0,7 in controls (difference of means: 0.7; $p < 0.001$); economic stress was reported in 70% of cases (OR 3,5). Early detection within the PHC setting reached 65% and resulted in a 28% reduction in anxiety levels. **Conclusions:** prenatal congenital heart defects were found to correlate with modifiable risk factors and were associated with disruptions in family cohesion. Primary Health Care facilitated early detection and support, thereby mitigating the negative impact in 30% of cases; the implementation of comprehensive programs is recommended.

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

INTRODUCTION

Congenital heart defects (CHD) are the most frequent structural anomaly at birth, with a global prevalence of 8 to 12 per 1,000 live births [1]. In Cuba, the National Perinatology Program has achieved high standards of prenatal detection; however, in peri-urban municipalities such as La Lisa, barriers derived from technological limitations in Primary Health Care (PHC) persist [2]. CHD not only affect fetal health but also generate psychosocial repercussions on family dynamics, such as stress, role alteration, and decreased family cohesion [3].

In the La Lisa municipality, the health areas of the Aleida Fernández, Cristóbal Labra, and Elpidio Berovides polyclinics present heterogeneous sociodemographic characteristics (central urban, peri-urban, and semi-rural), which could influence the prevalence of prenatal diagnosis of CHD and the subsequent family impact. However, no local studies have integrally analyzed maternal risk factors and repercussions on family dynamics from the perspective of Family and Community Medicine.

The objective of this study was to identify the risk factors associated with the prenatal diagnosis of congenital heart disease in pregnant women treated in three polyclinics in the La Lisa municipality, Cuba, during 2024.

METHODS

Study design

A descriptive, observational, cross-sectional study was conducted in the context of Primary Health Care (PHC) in the La Lisa municipality, Havana, Cuba, between February and August 2024.

Population and sample

The population consisted of 750 pregnant women treated in 18 family medical offices belonging to three polyclinics: Aleida Fernández (6 offices), Cristóbal Labra (8 offices), and Elpidio Berovides (4 offices).

A sample size of 360 participants was calculated considering a sampling error of 5%, a confidence level of 95%, and an expected prevalence of congenital heart defects of 9% according to previous national reports [2].

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

Sampling was proportional stratified by polyclinic and gestational trimester (first trimester: 40%, second: 35%, third: 25%), with simple random selection within each stratum.

Eligibility criteria

Inclusion criteria: Pregnant women with maternal age ≥18 years, who had at least one prenatal ultrasound performed at the PHC level.

Exclusion criteria: Multiple pregnancy, presence of non-cardiac congenital malformations prenatally diagnosed, refusal to participate through informed rejection.

A total of 375 pregnant women were selected, of which 360 agreed to participate (response rate of 96%).

Study variables

Independent variables (maternal risk factors): Maternal age (>35 years vs. ≤35 years), active smoking (≥5 cigarettes/day), parental consanguinity (2nd degree), gestational diabetes (clinical diagnosis according to MINSAP criteria), and pregestational body mass index (underweight: <18.5 kg/m², normal: 18.5-24.9 kg/m², overweight/obesity: ≥25 kg/m²).

Dependent variables: Prevalence of prenatally detected congenital heart defects (yes/no). Global score and subdomains of the Family Assessment Device (FAD) Scale (range 1-4; score >2.5 indicates family dysfunction).

Instruments and procedures

The following instruments were applied by previously trained nursing staff during routine prenatal consultations:

- Semi-structured survey (adapted from WHO Prenatal Risk Assessment): 35 items that explored maternal risk factors. It presented adequate internal consistency (Cronbach's α = 0.85).
- Family Assessment Device (FAD) Scale, short Spanish version: 12 items that evaluate cohesion, communication, problem-solving, and parental roles (α = 0.89).
- State-Trait Anxiety Inventory (STAI): 40 self-assessment items (score 1 to 4), applied before and after the psychological support intervention.

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

The diagnosis of congenital heart disease was made using fetal cardiac Doppler ultrasound in PHC, using Mindray portable equipment, with a reported sensitivity of 87% for major CHD. Cases with ultrasound suspicion were referred to the Pediatric Cardiology service of the William Soler University Pediatric Hospital for diagnostic confirmation.

Ethical aspects

The study was approved by the Research Ethics Committee of the La Lisa Municipality (Resolution No. 12/2024). Written informed consent was obtained from all participants. Anonymity was guaranteed through numerical codes and restricted access to the database. The design minimized risks and all participants received free psychological support at their office, regardless of the diagnostic result. The research was conducted in accordance with the principles of the Declaration of Helsinki (2013).

Statistical analysis

Data were processed using SPSS version 26 (IBM Corp., Armonk, NY, USA). Quantitative variables were summarized as mean ± standard deviation (SD) and qualitative variables as absolute frequencies and percentages.

To evaluate associations:

1. **Continuous variables with normal distribution:** Pearson correlation coefficient (r).
2. **Categorical variables:** chi-square test of independence and odds ratio (OR) with 95% confidence interval (95% CI).

Bonferroni correction was applied for post-hoc multiple comparisons. A p-value < 0.05 was considered significant.

RESULTS

Table 1 presents the maternal risk factors associated with the presence of prenatally diagnosed congenital heart defects (CHD), as well as the distribution of CHD prevalence by polyclinic. Risk factors are ordered from highest to lowest odds ratio (OR). Among the factors evaluated, maternal age >35 years showed the strongest association (OR = 3.1; 95% CI: 1.8-5.4; p = 0.001), followed by parental consanguinity (OR = 2.9; 95% CI: 1.6-5.2; p = 0.005).

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

Regarding geographic distribution, the Elpidio Berovides Polyclinic (semi-rural area) presented the highest prevalence of CHD (11%).

Table 1. Maternal risk factors and prevalence of congenital heart defects, with distribution by polyclinic (n=360)

Risk factor / Indicato	n	%	OR (IC 95%)	p-value	Aleida Fernández (n=119)	Cristóbal Labra (n=162)	Elpidio Berovides (n=79)
Risk factors							
Maternal age >35 years	101	28,1	3,1 (1,8-5,4)	0,001	25%	30%	32%
Active smokin	65	18,1	2,6 (1,4-4,8)	0,003	15%	17%	22%
Parental consanguinity	36	10,0	2,9 (1,6-5,2)	0,005	8%	11%	13%
Gestational diabetes	43	11,9	2,1 (1,1-4,0)	0,032	10%	13%	14%
Low pregestational	52	14,4	1,8 (1,0-3,3)	0,048	12%	15%	18%
CHD prevalence	33	9,2	—	—	8%	10%	11%

Source: Own survey, 2024. **Note:** CHD = congenital heart defects; BMI = body mass index; OR = odds ratio; CI = confidence interval; n = absolute frequency; % = valid percentage. Percentages by polyclinic correspond to the proportion within each center. Difference between polyclinics for CHD prevalence: p = 0.03 (chi-square test).

Table 2 shows the Pearson correlations. Family unemployment presented the highest correlation with family dysfunction (r = 0.58), followed by smoking with CHD severity (r = 0.45).

Table 2. Pearson correlations between risk factors, CHD, and family dynamics (n=360)

Variables	r (Pearson)	p-value	Interpretation (Cohen)
Maternal age and CHD	0,41	<0,001	Moderada

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

risk			
Smoking and CHD severity	0,45	<0,001	Moderada
Gestational diabetes and FAD	0,35	0,004	Moderada
Family unemployment and FAD score	0,58	<0,001	Grande
Consanguinity and family cohesion (FAD)	-0,38	0,001	Moderada

Source: Own analysis, 2024. **Note:** FAD = Family Assessment Device; r > 0.4 moderate; r > 0.5 large; negative values indicate inverse correlation.

Table 3 compares family dynamics between cases and controls. Cases with CHD exceeded the dysfunction cut-off point (>2.5) in cohesion, problem-solving, and parental roles, while controls remained below in all subdomains.

Table 3. Family repercussions by FAD subdomain in CHD cases (n=33) vs. controls (n=327)

FAD Subdomain	CHD Cases (n=33)	Controls (n=327)	Difference of means	p-value
Family cohesion	2,8 ± 0,9	1,7 ± 0,6	1,1	<0,001
Communication	2,5 ± 0,8	1,9 ± 0,7	0,6	0,002
Problem-solving	2,7 ± 1,0	2,0 ± 0,8	0,7	<0,001
Parental roles	2,9 ± 0,9	2,1 ± 0,7	0,8	<0,001

Source: Family Assessment Device (FAD) Scale, 2024.**Note:** Score range 1-4; >2.5 indicates family dysfunction. Data presented as mean ± standard deviation.

DISCUSSION

The prevalence of prenatally detected congenital heart defects (CHD) in the La Lisa municipality (9.2%) is consistent with Cuban national reports and with World Health Organization estimates [1,2]. This finding confirms that, despite technological limitations in Primary Health Care (PHC), the National Perinatology Program maintains detection standards comparable to those in the region [4].

The identified risk factors —maternal age >35 years (OR 3.1), parental consanguinity (OR 2.9), and active smoking (OR 2.6)— are consistent with those reported in the international literature. The magnitude of the association for advanced maternal age in this study (OR 3.1) is higher than that reported in some recent meta-analyses,

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

where evidence is not conclusive [5], which could reflect differences in the distribution of other unmeasured risk factors in the population.

Active smoking, with a moderate correlation with CHD severity (r = 0.45), stands out as a key modifiable risk factor. In La Lisa, the prevalence of smoking in pregnant women (18%) exceeds the last available national figure of 12% [2], which underlines the urgency of strengthening anti-smoking interventions within the Family Doctor and Nurse Program (FDNP), especially in the most vulnerable areas such as Elpidio Berovides.

The variation in CHD prevalence by polyclinic (Elpidio Berovides 11% vs. Aleida Fernández 8%; p = 0.03) suggests a possible influence of social and environmental determinants typical of semi-rural areas. This finding aligns with regional analyses from the Pan American Health Organization that document perinatal health inequalities associated with socioeconomic level [4], as well as with Cuban studies that confirm the incidence of social determinants on infant mortality [6].

The impact of prenatal diagnosis of CHD on family dynamics was significant. The overall mean FAD score in cases (2.6 ± 0.9) exceeded the family dysfunction threshold (>2.5), while controls remained below (1.9 ± 0.7). This difference (0.7 points) is clinically relevant and consistent with the literature documenting that families with fewer psychosocial resources and lower levels of support present a higher risk of psychological distress and lower well-being over time [7].

The most affected subdomains were parental roles (2.9 ± 0.9) and family cohesion (2.8 ± 0.9), which agrees with previous research indicating that prognostic uncertainty and medical care demands alter the distribution of responsibilities within the home (7). Economic stress, reported by 70% of families with CHD (OR 3.5), emerges as an amplifying factor of dysfunction, especially in a context of social inequalities and resource limitations as currently in Cuba [8].

Despite barriers, Cuban PHC proved to be a fundamental pillar. Early detection in the first trimester (65% of cases) allowed timely referral to the tertiary level in 75% of pregnant women, and integrated psychological support in the offices reduced prenatal anxiety by 28% (measured with the STAI inventory). These results validate the effectiveness of the Cuban comprehensive care model, which

MedEst. 2026| Vol. 6: e523
ISSN: 2789-7567 | RNPS: 2524

combines ultrasound screening with psychosocial support, mitigating the negative impact in approximately one third of cases [9].

From a community perspective, the differences observed between polyclinics suggest the need for differentiated interventions: in urban areas such as Aleida Fernández, strengthen prenatal education and risk factor detection; in semi-rural areas such as Elpidio Berovides, prioritize psychosocial support and family resilience programs.

This study has several limitations that should be considered. First, the cross-sectional design prevents establishing causal relationships between risk factors and CHD, as well as evaluating the evolution of family dynamics after birth. Second, stratified sampling, although representative, could underestimate prevalence in sectors with greater logistical barriers (for example, pregnant women who do not attend regular prenatal check-ups). Third, the 87% sensitivity of Doppler ultrasound in PHC, although acceptable, implies that some mild cases of CHD may not have been detected (false negatives). Finally, the use of Pearson correlation for ordinal variables (smoking and CHD severity) constitutes a statistical limitation; analysis with Spearman correlation could offer more precise estimates.

Despite these limitations, the findings are applicable to other Cuban municipalities with similar sociodemographic characteristics and to middle-income contexts where PHC is the first level of contact. The combination of ultrasound screening with validated instruments such as the FAD scale and STAI can be replicated in other health areas. It is recommended to expand Doppler ultrasound coverage and design family resilience programs in the offices to mitigate the detected dysfunctions, following multidimensional psychosocial care models validated in pediatric populations with CHD (10), in line with the guidelines of the Cuban Maternal and Child Health Care Program [11].

CONCLUSIONS

In summary, the prenatal diagnosis of congenital heart defects in La Lisa was associated with modifiable risk factors (low pregestational BMI, smoking) and with significant family dysfunction, with geographical variations that concentrated the greatest burden in the semi-rural polyclinic (Elpidio Berovides). Cuban Primary Health Care proved to be effective in early detection and reduction of prenatal anxiety, although challenges related to technological limitations and socioeconomic inequalities persist. Differentiated community

interventions are recommended to strengthen prenatal education in urban areas and family resilience programs in rural and peri-urban areas.

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HPH: Conceptualization, data curation, formal analysis, investigation, project administration, supervision, visualization, writing, original draft.

CONFLICTS OF INTEREST

The authors declare that there are no conflicts of interest.

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The authors declare that artificial intelligence was not used in the writing of this manuscript.