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Right Aortic Arch in Pediatric: Experience of a Center

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Abstract

Background: Congenital aortic arch anomalies encompass various malformations, which can range from asymptomatic to presenting with severe respiratory or gastrointestinal issues. These anomalies may occur alone or alongside other congenital heart defects. Imaging is crucial for identifying these conditions and guiding accurate preoperative decisions.

Objectives: This study aimed to assess the frequency of these abnormalities in patients with a right aortic arch, explore the reasons for their referral, and evaluate patient outcomes.

Methods: This cross-sectional study was conducted on children under the age of 20 who visited the Congenital Heart Diseases Clinic at Imam Reza Hospital, Iran. We collected data from 277 records of patients with right aortic arch from 2002 to 2022. Data were gathered using a checklist, including echocardiographic results, angiography findings, and other diagnostic modalities for the patients. Statistical analysis was performed using IBM SPSS software, Version 25.

Results: In this study, 118 (29.8%) patients had heterotaxia, of which 106 (26.8%) had dextrocardia and were excluded [82 (20.7%) with situs inversus, 21 (5.3%) with situs solitus, and 15 (3.7%) with situs ambiguous]. Among the 277 patients with a right aortic arch, 7 (2.3%) had an aberrant left subclavian artery branch. There were 141 (50.9%) female patients. The trachea and esophagus were compressed. Respiratory symptoms were commonly present in patients with a compressive right arch and aberrant left subclavian artery (Kommerell's diverticulum), which showed a significant relationship. Congenital major heart anomalies were found in 261 (94.2%) patients, with the most common being right obstructive lesions related to tetralogy of fallot (TOF), seen in 115 (41.5%) patients. Complex congenital heart disease was present in 75 (27.1%) patients. Septal defects were observed in 44 (15.9%) patients, including ventricular septal defect in 39 (14.0%). Nine (3.2%) patients had left obstructive lesions, including coarctation of the aorta. Arterial anomalies, such as patent ductus arteriosus, were present in 14 (5.0%) patients. Sixteen patients had normal heart structures or minor cardiac anomalies. The most common non-cardiac abnormality was musculoskeletal. Finally, 189 (68.2%) patients underwent surgery, and 16 (5.8%) died.

Conclusions: More detailed examinations during patient visits, attention to clinical symptoms, and modern imaging can hasten the identification of cardiac abnormalities in children, which plays an important role in the diagnosis and prognosis of patients.

Keywords: Heart Defects, Aortic Arch Syndromes, Pediatrics

1. Background

Aortic arch abnormalities are a diverse group of congenital anatomical defects of the main artery. The treatment process and symptoms of the disease vary depending on whether they are accompanied by other congenital cardiovascular, respiratory, and digestive defects (1). Congenital abnormalities of the aortic arch refer to abnormal embryologic development of the aorta and its branches, which can be characterized as interrupted, right-sided, left-sided, or double in configuration, depending on the course and position of the aortic arch (2).

In infants, aortic anomalies can lead to clinical signs such as cyanosis, dysphagia from tracheal and esophageal compression, and respiratory distress (3). A

right aortic arch (RAA), which courses to the right of the trachea in contrast to a normal left aortic arch, is due to abnormal regression of the primordial aortic arch system during embryogenesis. This anomaly may be associated with congenital heart defects (CHD) as well as extracardiac anomalies. The RAA is a rare anomaly with an incidence of 0.1%, and patients typically develop symptoms between 1 and 6 months of age (4, 5). It is often associated with other cardiovascular abnormalities, including tricuspid atresia, tetralogy of fallot (TOF), truncus arteriosus, and transposition of the great arteries with pulmonary valve stenosis (2).

Kommerell's diverticulum, a congenital malformation with a prevalence of 0.04% to 0.4%, was first described by Kommerell in 1936 as an aneurysmal dilation at the base of the left subclavian artery. This dilation can enlarge and cause tracheal and esophageal compression, leading to symptoms such as dysphagia and respiratory distress (5, 6). Radiologists, vascular surgeons, and thoracic surgeons must recognize Kommerell's diverticulum on thoracic CT angiography (7). The embryological development of the RAA involves complex interactions of the pharyngeal arches and the dorsal aortas, leading to various types of RAA configurations (8). As imaging plays a significant role in diagnosing these abnormalities, techniques such as fetal echocardiography, multi-detector three-dimensional CT, and three-dimensional Magnetic Resonance Angiography provide detailed assessments of tracheoesophageal anatomy (9, 10).

2. Objectives

This study aimed to investigate the frequency and outcomes of RAA in children admitted to Imam Reza Hospital in Mashhad, Iran.

3. Methods

This cross-sectional study was conducted on pediatric patients under the age of 20 who were referred to the Congenital Heart Diseases Clinic of Imam Reza Hospital in Mashhad, Iran. The data collection spanned two decades, from 2002 to 2022, providing a robust dataset for analyzing long-term trends and outcomes in congenital heart disease.

All patients diagnosed with aortic arch anomalies or other heart defects were assessed. Using the census method, every case that met the criteria was included in the study to ensure a thorough representation of the patient population. The study utilized a variety of diagnostic tools, including epidemiological assessments, echocardiographic evaluations, angiography, and other diagnostic modalities. Additionally, surgical treatments were performed as needed, and all relevant information was meticulously collected by a trained expert using a standardized form. Compressive symptoms were noted as gastrointestinal (e.g., dysphagia) and respiratory (e.g., noisy respiration, chronic cough, recurrent upper respiratory infections).

This study also investigated patients for non-cardiac anomalies, including dermal, gastrointestinal, urology, musculoskeletal, endocrine, ocular, eye, ears, nose, throat (ENT), pharynx, and neuropsychiatric anomalies. We first examined the types of major and minor cardiac anomalies. The major anomalies included:

- Septal defects: Ventricular septal defect (VSD), atrial septal defect (ASD), atrioventricular septal defect (AVSD)

- Right-sided lesions: Tetralogy of fallot (TOF), pulmonary stenosis (PS)

- Left-sided lesions: Aortic stenosis (AS), coarctation of the aorta (COA)

- Arterial abnormalities: Patent ductus arteriosus (PDA)

- Mitral valve abnormalities: Congenital mitral stenosis or mitral regurgitation

- Tricuspid valve abnormalities: Ebstein anomaly

We separately expressed the frequency of these anomalies and their relationship with the RAA anomaly. Patients with normal echocardiograms or heart structures, or those with minor and unimportant findings (i.e., those that do not require intervention or follow-up), were categorized into normal and minor groups. These included conditions such as PFO, FMV without MR or trace MR, and LSVC. In contrast, cases with structural defects were considered abnormal and important (major anomalies).

"Complex" refers to situations where there are more than three main categories of associated problems, or when these are accompanied by conditions such as cardiopathies, TGA, malposed great arteries, DORV, tricuspid atresia, and single ventricle.

Patients were classified into five groups based on their referral causes: Cardiac, respiratory, gastrointestinal, cases referred for consultation from other colleagues, and an "others" group that included syndromic appearances, Down syndrome appearance, abnormal chest X-ray, cleft lip, fetal echo findings, failure to thrive (FTT), imperforate anus, and ruling out vascular anomalies.

Once the data were collected, they underwent thorough statistical analysis to identify significant trends and outcomes that could provide meaningful insights into the study's objectives. The statistical analysis was performed using IBM SPSS software (Version 25, SPSS Inc., Chicago, IL, USA), a widely used tool for managing and analyzing large datasets. The significance level for all statistical tests was set at ≤ 0.05, meaning that any P-value below this threshold would be considered statistically significant. Descriptive statistics, including mean, standard deviation, number, and percentage, were used to summarize the data and provide an overview of the sample characteristics. To assess the relationships between categorical variables, a chi-square test was applied.

4. Results

This study was conducted on 277 patients with a RAA. Among these patients, 7 (2.3%) had a branch of the left subclavian artery, which consequently developed Kommerell's diverticulum. A total of 141 (50.9%) patients were girls. Of the patients, 86.6% had a normal birth weight. Regarding the weight at the time of diagnosis and treatment, 31.4% of patients weighed between 10-15 kg. In the analysis of birth weight and patients who underwent surgery, 162 patients (85.7%) had a normal birth weight, 4 patients (2.1%) had above-normal weight, and 23 patients (12.2%) had below-normal birth weight. No significant relationship was found between birth weight and surgical cases. Among the patients who underwent surgery, 106 (56.1%) were under one year old, 23 (12.2%) were aged 1 to 2 years, 21 (11.1%) were aged 3 to 5 years, 25 (13.2%) were aged 6 to 12 years, and 14 (7.4%) were over 12 years old. No significant associations were found between age groups and surgical cases (P = 0.552). Additionally, 12 patients (75%) who expired were under one year of age.

Among all patients, 7 exhibited compressive symptoms and developed Kommerell's diverticulum. All of these patients underwent surgery, though no significant relationship was found between compressive symptoms and surgical intervention (P = 0.067). The outcomes for these 7 patients were stable, with no association between compressive symptoms and overall outcomes (P = 0.722). Of the 7 patients, 6 had respiratory symptoms and 1 had gastrointestinal symptoms. Respiratory tract symptoms showed a significant association with RAA anomalies and Kommerell's diverticulum (P < 0.001), as did gastrointestinal symptoms (P < 0.001).

Additionally, 75 patients (27.0%) had concurrent non-cardiac anomalies. The most common non-cardiac anomaly was musculoskeletal, present in 21 patients (7.5%), followed by neuropsychiatric anomalies in 14 patients (5.1%) and endocrine anomalies in 12 patients (4.3%). Other notable anomalies included urological (10 patients, 3.6%), gastrointestinal (7 patients, 2.5%), and both eye and ENT anomalies (4 patients each, 1.4%). Additionally, 3 patients (1.0%) had concurrent non-cardiac anomalies. The relationship between the non-cardiac anomalies and the outcome of the patients showed that endocrine anomalies had a highly significant P-value of < 0.0001, while musculoskeletal (0.72) and gastrointestinal (0.72) anomalies showed no significant association. Other anomalies, such as neuropsychiatric (0.32) and ENT (0.83), also demonstrated non-significant results (Table 1).

The relationship between non-cardiac anomalies and surgical intervention showed that significantly all patients with urological anomalies required surgery (P = 0.034). Other anomalies, including musculoskeletal, neuropsychiatric, and endocrine, displayed non-significant P-values (0.87, 0.74, and 0.99, respectively). Gastrointestinal and eye anomalies also showed non-significant results, with P-values of 0.10 and 0.99, respectively (Table 2).

Patients with cardiac anomalies were divided into two groups: Major and minor. Out of the total patients, 261 (94.2%) had major cardiac anomalies, which included 119 patients (42.9%) with right-sided anomalies, 75 patients (27.1%) with complex anomalies, 44 patients (15.9%) with septal defects, 14 patients (5.0%) with arterial abnormalities, and 9 patients (3.2%) with left-sided

Table 1. The Relationship Between the Non-cardiac Anomalies and the Outcome of the Patients

Anomaly	No. (%)	Patients' Outcome			P-Value [a]
		Stable	Missed Follow-up	Expired	
Musculoskeletal	21 (28.3)				0.72
Yes		20 (95.2)	0	1 (4.8)	
No		234 (91.4)	7 (2.7)	15 (5.9)	
Neuropsychiatric	14 (18.9)				0.32
Yes		12 (85.7)	0	2 (14.3)	
No		242 (92)	7 (2.7)	14 (5.3)	
Endocrine	12 (16.2)				0.0001
Yes		6 (50)	2 (16.7)	4 (33.3)	
No		248 (93.6)	5 (1.9)	12 (4.5)	
Urology	10 (13.5)				0.62
Yes		10 (100)	0	0	
No		244 (91.4)	7 (2.6)	16 (6)	
Gastrointestinal	7 (9.4)				0.72
Yes		7 (100)	0	0	
No		247 (91.5)	7 (2.6)	16 (5.9)	
Eye	4 (5.4)				0.24
Yes		3 (75)	0	1 (25)	
No		251 (91.9)	7 (2.6)	15 (5.5)	
ENT	4 (5.4)				0.83
Yes		4 (100)	0	0	
No		250 (91.6)	7 (2.6)	16 (5.9)	
Urology and musculoskeletal	3 (4.0)				0.87
Yes		3 (100)	0	0	
No		251 (91.6)	7 (2.6)	16 (5.8)	

Abbreviations: ENT, ear, nose and throat.

[a] Chi-square test.

anomalies. The most common major cardiac anomaly was TOF, observed in 115 patients (41.5%). Additionally, there were 16 patients (5.8%) with minor cardiac anomalies or normal findings. The outcomes for patients with septal defects showed a P-value of 0.19, indicating no significant association. In contrast, left-sided lesions had a significant P-value of 0.001, while complex CHD also showed significant results with a P-value of 0.004. Other anomalies, including arterial abnormalities and minor anomalies, had non-significant P-values of 0.51 and 0.46, respectively (Table 3).

It is important to note that major cardiac anomalies also include abnormalities of the mitral and tricuspid valves, for which we had no patients in these groups; therefore, they were excluded from the table. Additionally, some groups contained patients with concurrent anomalies, which are referenced in the table of major cardiac anomalies presented earlier in the article.

Assessing the relationship between patients' major and minor cardiac anomalies and the surgery revealed a significant association for arterial abnormalities with a P-value of 0.04 and right-sided lesions with a P-value of 0.01. Other anomalies, including septal defects (0.15), left-sided lesions (0.14), complex congenital heart disease (0.13), and minor anomalies (0.54), showed no significant associations with surgical outcomes. These findings indicated that certain cardiac anomalies are more likely to require surgical intervention than others (Table 4).

In terms of patient referrals, cardiac causes accounted for 166 patients (59.9%), respiratory causes for 19 patients (6.9%), gastrointestinal causes for 1 patient (0.4%), consultations for 61 patients (22%), and other causes for 30 patients (10.8%). There was no relationship

Table 2. The Relationship Between Patients' Non-cardiac Anomalies and the Surgery [a]

Anomaly	Surgery		P-Value [b]
	Yes	No	
Musculoskeletal			0.87
Yes	14 (66.7)	7 (33.3)	
No	175 (68.4)	81 (31.6)	
Neuropsychiatric			0.74
Yes	9 (64.3)	5 (35.7)	
No	180 (68.4)	83 (31.6)	
Endocrine			0.99
Yes	8 (66.7)	4 (33.3)	
No	181 (68.3)	84 (31.7)	
Urology			0.034
Yes	10 (100)	0	
No	179 (67)	88 (33)	
Gastrointestinal			0.10
Yes	7 (100)	0	
No	182 (67.4)	88 (32.6)	
Eye			0.99
Yes	3 (75)	1 (25)	
No	186 (68.1)	87 (31.9)	
ENT			0.59
Yes	2 (50)	2 (50)	
No	187 (68.5)	86 (31.5)	
Urology and musculoskeletal			0.55
Yes	3 (100)	0	
No	186 (67.9)	88 (32.1)	

Abbreviations: ENT, ear, nose and throat.

[a] Values are expressed as No. (%).

[b] Chi-square test.

between referral reasons and surgical cases (P = 0.899). Additionally, 155 patients (93.4%) referred due to cardiac issues had stable outcomes, but no correlation was found between referral reasons and patient outcomes (P = 0.530).

Table 5 outlines the frequency of cardiac abnormalities associated with RAA in patients, revealing that a majority had major anomalies, including septal defects and TOF. It also indicates that minor anomalies were present in a smaller percentage of the cohort, highlighting the diverse nature of cardiac conditions linked to the RAA.

Assessing the outcomes of patients with RAA anomalies and their surgical interventions, a significant majority (91.7%) had stable outcomes after treatment. A small percentage (2.5%) were lost to follow-up, while 5.8% expired. Regarding surgical intervention, 68.2% of patients underwent surgery, demonstrating a substantial inclination toward surgical management in this population, with 31.8% not requiring surgical intervention. This table underscores the effectiveness of surgical approaches in improving patient stability.

5. Discussion

Congenital heart diseases represent a broad spectrum of structural and functional abnormalities that affect infants, and they vary significantly in type and severity. Right aortic arch anomalies are among these congenital defects, occurring either as isolated anomalies or in association with other congenital heart conditions, particularly TOF. This study aimed to examine the frequency, associated anomalies, and outcomes in pediatric patients with RAA, contributing to the growing body of literature that seeks to enhance

Table 3. The Relationship Between the Major and Minor Cardiac Anomalies and the Outcome of the Patients [a]

Anomaly	Patients' Outcome			P-Value [b]
	Stable	Missed Follow-up	Expired	
Septal defect				0.19
Yes	43 (97.7)	1 (2.3)	0	
No	211 (90.6)	6 (2.6)	16 (6.9)	
Arterial ABN				0.51
Yes	14 (100)	0	0	
No	240 (91.3)	7 (2.7)	16 (6.1)	
Left ABN				0.001
Yes	7 (77.8)	2 (22.2)	0	
No	247 (92.2)	5 (1.9)	16 (6)	
Right ABN				0.55
Yes	116 (93.5)	2 (1.6)	6 (4.8)	
No	138 (90.2)	5 (3.3)	10 (6.5)	
Complex CHD				0.004
Yes	63 (84)	2 (2.7)	10 (13.3)	
No	191 (94.5)	5 (2.5)	6 (3)	
Minor anomalies				0.46
Yes	16 (100)	0	0	
No	238 (91.2)	7 (2.7)	16 (6.1)	

Abbreviations: CHD, congenital heart disease; ABN, abnormality.

[a] Values are expressed as No. (%).

[b] Chi-square test.

understanding of these rare congenital heart conditions.

A systematic review identified 99 patients who underwent surgical or endovascular intervention for RAA aneurysm or dissection. Among these, 88 patients had an aberrant left subclavian artery, with only 11 presenting a mirror-image RAA. Notably, the most common pathology was aneurysm related to Kommerell's diverticulum, observed in over 50% of cases (11). In our study, 7 out of 277 patients were diagnosed with Kommerell's diverticulum, all of whom underwent surgery, further supporting the rarity of this condition.

A retrospective analysis involving 92 fetuses diagnosed with RAA confirmed that TOF was the most frequently associated congenital anomaly. This emphasizes the strong association between RAA and TOF, indicating that these conditions often coexist. Similarly, in our study, 7 patients were identified with an aberrant left subclavian artery, a known vascular anomaly that can arise alongside RAA. Notably, 6 of these patients presented with respiratory symptoms, likely due to the compression of the trachea or esophagus by the aberrant vascular structures, leading to clinical manifestations such as stridor, difficulty breathing, or feeding issues. All these cases required surgical intervention to alleviate symptoms and prevent long-term complications (12).

These findings underscore the critical need for comprehensive clinical assessments during routine patient visits, especially in cases where congenital heart anomalies like RAA are suspected or diagnosed prenatally. Early recognition of symptoms, particularly respiratory distress linked to vascular anomalies, is essential for timely surgical planning. This proactive approach not only improves diagnostic accuracy but also enables timely interventions that can significantly reduce morbidity and enhance the quality of life for affected infants. Therefore, integrating detailed prenatal imaging and postnatal evaluations can be crucial in managing such complex congenital conditions effectively.

A study conducted between 2006 and 2017, which reviewed 36 patients with RAA, identified TOF as the most commonly associated anomaly, underscoring the

Table 4. The Relationship Between Patients' Major and Minor Cardiac Anomalies and the Surgery [a]

Anomaly	Surgery		P-Value [b]
	Yes	No	
Septal defect			0.15
Yes	26 (59.1)	18 (40.9)	
No	163 (70)	70 (30)	
Arterial ABN			0.04
Yes	13 (92.9)	1 (7.1)	
No	176 (66.9)	87 (33.1)	
Left ABN			0.14
Yes	4 (44.4)	5 (55.6)	
No	185 (69)	83 (31)	
Right ABN			0.01
Yes	94 (75.8)	30 (24.2)	
No	95 (62.1)	58 (37.9)	
Complex CHD			0.13
Yes	46 (61.3)	29 (38.7)	
No	143 (70.8)	59 (29.2)	
Minor anomalies			0.54
Yes	12 (75)	4 (25)	
No	177 (67.8)	84 (32.2)	

Abbreviations: PDA, patent ductus arteriosus; CHD, congenital heart disease; ABN, abnormality.

[a] Values are expressed as No. (%).

[b] Chi-square or fisher exact test.

strong correlation between RAA and TOF. This study not only documented anatomical associations but also included genetic testing for 16 of the cases, providing a more comprehensive analysis of potential genetic factors. Among these, two patients were found to have a 22q11.2 microdeletion, a genetic abnormality commonly linked to congenital heart defects, particularly TOF. Additionally, two cases involved trisomy 21, another chromosomal disorder associated with various congenital anomalies (13). The identification of these genetic abnormalities highlights the potential role of genetic factors in the etiology of RAA and its associated heart defects. Given these findings, incorporating genetic analysis in future studies could significantly enhance the understanding of the pathophysiological mechanisms underlying congenital heart diseases. It may also aid in early diagnosis, risk stratification, and tailored management strategies for affected patients, ultimately improving clinical outcomes.

A study evaluating 45 pregnancies with a prenatal diagnosis of RAA demonstrated that 82.2% of cases exhibited a U-shaped configuration, with a complete vascular ring observed in all instances. Additionally, 56.8% of these cases were associated with a RAA and Kommerell's diverticulum, indicating a relatively high prevalence in this cohort (4). Conversely, our study identified a significantly lower incidence of Kommerell's diverticulum at 2.3%. This discrepancy suggests that prenatal imaging techniques might play a crucial role in the early detection of vascular anomalies, potentially allowing for prompt medical or surgical management, thereby reducing the risk of complications associated with these congenital conditions.

Additionally, a study examining 71 cases of RAA identified a range of associated congenital anomalies, with TOF and pulmonary atresia being frequently observed. This finding highlights the strong association between RAA and complex CHD (14). Our study further supported these results, as TOF emerged as the most common cardiac abnormality in our cohort, emphasizing the consistency of this association across different populations. This reinforces the need for comprehensive prenatal screening in fetuses diagnosed

Table 5. The Relation Between Right Aortic Arch, with Associated Cardiac Abnormalities in Patients [a]

Variables	Right Aortic Arch
Major	261 (94.2)
Septal defected	44 (15.9)
AVSD	0 (0.0)
VSD	39 (14.07)
ASD	3 (1.08)
ASD + VSD	2 (0.7)
Arterial	14 (5.05)
PDA	7 (2.52)
PDA + VSD	2 (0.7
PDA + TOF	5 (1.8)
RT. side lesion	119 (42.9)
TOF	115 (41.5)
PS	4 (1.44)
LT. side lesion	9 (3.2)
COA	9 (3.2)
AS	0 (0.0)
MV abnormality	
Congenital MS or MR	0 (0.0)
TV abnormality	
Ebstein anomaly	0 (0.0)
Complex	75 (27.1)
Others	75 (27.1)
Minor	16 (5.8)

Abbreviations: AVSD, atrioventricular septal defect; VSD, ventricular septal defect, ASD, atrial septal defect; PDA, patent ductus arteriosus; TOF, tetralogy of fallot; PS, pulmonic stenosis; COA, coarctation of the aorta; AS, aortic stenosis; MV, mitral valve; TV, tricuspid valve.

[a] Values are expressed as No. (%).

with a RAA to identify concurrent anomalies, which may critically influence clinical management and surgical planning.

The strength of our study lies in its comprehensive duration of 20 years, conducted at a major referral hospital in eastern Iran, which enabled a diverse patient population for robust statistical analysis. Unlike many prior studies, which were mainly descriptive, our research provides a thorough examination of the relationships between cardiac abnormalities, demographic factors, surgical interventions, and patient mortality.

5.1. Conclusions

In conclusion, this study emphasizes the critical role of early detection of congenital heart anomalies using accessible diagnostic methods. Timely identification can significantly reduce complications and enhance treatment outcomes. By conducting detailed examinations, closely monitoring clinical symptoms, and employing advanced imaging techniques, healthcare providers can improve diagnostic accuracy and patient prognosis in pediatric populations. Future research should continue to explore genetic factors associated with CHD to further enhance understanding and management strategies.

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Footnotes

Authors' Contribution: B. B. and A. M. M. Sh.: Conceived and designed the analysis, collected the data,

contributed data or analysis tools, and wrote the draft of the paper; M. H. M. and M. A.: Conceived and designed the analysis, collected the data, and revised the paper; H. M. M. Sh.: Conceived the analysis, contributed data or analysis tools, and revised the paper. All authors revised the study and approved it.

Conflict of Interests Statement: The fourth and fifth authors are son and father.

Data Availability: All data generated or analyzed during this study are included in this published article.

Ethical Approval: All experiments were performed following relevant guidelines and regulations of the Declaration of Helsinki. The Ethical approval was obtained from the ethics committee of Mashhad University of Medical Sciences (ethical code: IR.MUMS.MEDICAL.REC.1399.083).

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