

# Interrupted aortic arch – case report

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## ABSTRACT

Interrupted aortic arch is a rare ductal-dependent congenital heart malformation, accounting for 1.5% of all MCC, 2 in 100,000 cases of live newborns are diagnosed with IAA. We present the case of a 3-month-old infant who came to our clinic on the recommendation of the family doctor. On clinical examination, he presented an affected general state, perioronasal cyanosis during moments of agitation, grade III/6 systolic murmur present throughout the precordial area, heart rate = 160 beats/min, respiratory rate = 45 breaths/min, SaO<sub>2</sub> = 86 – 91% in atmospheric air, liver 4 cm below the costal rim. Echocardiography revealed a complete ductal-dependent congenital heart malformation, interrupted aortic arch, persistence of ductus arteriosus, wide muscular ventricular septal defect, hypoplasia of the ascending aorta. The angio-CT examination confirmed the diagnoses from the echocardiography. During the genetic examination, the suspicion of DiGeorge syndrome was raised, which is why MLPA for microdeletions was required. The first therapeutic act performed was represented by maintaining the patency of the arterial channel. The patient was transferred to a clinic in Italy for the surgical intervention.

**Keywords:** Interrupted aortic arch, congenital heart malformation, DiGeorge syndrome

## INTRODUCTION

Interrupted aortic arch is a rare congenital ductal-dependent cardiac malformation, accounting for 1.5% of all MCC, 2 in 100,000 cases of live newborns are diagnosed with IAA [1,2]. There is a partial or total discontinuity of the lumen aortic between the ascending aorta and the descending aorta, if the discontinuity is partial, the lumen appears as a band of fibrous tissue, but with hemodynamic effects similar to the total discontinuity of the aortic lumen [1,3]. This aortic arch malformation has a mortality of approximately 90% in the absence of prompt surgical intervention [1,2,3,4].

Steidele, describes for the first time the interrupted aortic arch in 1778, and Celoria and Palton classify the interrupted aortic arch according to the place where the discontinuity is on the path of the aortic arch into three types, type A, Type B, type C [3,5,6,7,8]. According to studies, type B is the most common type of interrupted aortic arch, with a prevalence of 52% - 90% [3].

The association of AI with other cardiac malformations is common, the most common being the interventricular septal defect (70% – 80%) [3,9].

Genetically, IAA associates in a percentage of 50% of cases the 22q11.2 deletion, this deletion has been associated in a large percentage (75%–85%) with congenital heart malformations. The 22q11.2 deletion is also present in patients diagnosed with DiGeorge Syndrome, a genetic condition that in 50% of cases is also found with IAA [1,2,3].

The clinical picture can be initially poor, patients can be asymptomatic until the moment the atrial canal closes, most often in the first 72 hours after birth, the onset of symptoms overlaps with the closure of the atrial canal, they vary from signs of heart failure to cardiogenic shock and death in the absence of medical intervention [3,10,11].

During the clinical examination, we will be able to observe the bulging of the precordial region with ample pulsations of the right ventricle, on listening we will detect a galloping rhythm, with a systolic

murmur grade II -III/ 6 basal or holosystolic murmur and 2 doubled noises [3,10,11].

The diagnosis can be made from intrauterine life, the long axis of the heart with the emergence of the aorta can help us highlight the intrauterine aortic arch, both in the three-vessel section with the trachea and in the parasagittal section, a portion of the aortic arch can be visualized, helped by Doppler color [12,13,14].

Postnatally, clinical examination together with paraclinical investigations will establish the diagnosis. Echocardiography is the preferred investigation for establishing the diagnosis, being a non-invasive maneuver, it highlights and describes the location of the interruption of the aortic arch.

The therapeutic management is divided into three moments defined by the surgical act; thus, we will have a preoperative, operative and postoperative approach.

Essential immediately after the diagnosis of IAA is the start of prostaglandin E1 (PGE1) therapy, with the role of maintaining or reopening the arterial channel, followed by hemodynamic stabilization [3,7,8]. Medical advances have shortened the appropriate time for surgery from 6 weeks to the first week of life. According to the studies, it is preferable to solve the IAA in a single surgical act, a method associated with fewer reinterventions [15,16,17].

We present to you a special case of interrupted aortic arch, a duct dependent congenital heart malformation, in an infant, which associates wide muscular interventricular septal defect, hypoplasia of the ascending aorta, pulmonary arterial hypertension, Ross class IV heart failure, with DiGeorge syndrome in observation.

The written informed consent was obtained from the patient's mother prior to publication of this case.

## CASE PRESENTATION

We present the case of a 3-month-old infant, who was hospitalized for the first time in the Cardiology Clinic on the recommendation of the family doctor.

During the clinical examination, he presented a general affected state, peri oronasal cyanosis during moments of agitation, grade III/6 systolic murmur present in the entire precordial area, heart rate = 160 beats/min, respiratory rate = 45 breaths/minute, SaO<sub>2</sub> = 86 – 91% in atmospheric air, liver 4 cm below the costal margin. He was fed by gavage with good digestive tolerance.

### Paraclinical examinations

The echocardiography performed at admission (25.08.2021) revealed a wide muscular ventricular septal defect (5 mm), with a left-right shunt present,

the trunk of the pulmonary artery significantly dilated, pulmonary artery – 14 mm, Z score = + 4.38, with a pressure gradient of 7 mmHg, dilated left branch of the pulmonary artery – Z score = + 3.69, dilated left heart, ascending aorta and course of the aorta were hypoplastic and interrupted, descending aorta with apparent origin from the left branch of the pulmonary artery plus reduced caliber.

On 26.08.2021, a CT angiogram was performed, which confirmed the suspicion of an interrupted aortic arch, with the origin of the descending aorta from the arterial canal, which is still persistent with a caliber of 4 mm, the descending aorta with a caliber of 58 mm, the sub clavicular artery left with a caliber of 23 mm, ascending aorta with a caliber of 5 mm. The trunk of the pulmonary artery has an increased caliber - 117 mm, the branches of the pulmonary artery - right branch - 68 mm, left branch 90 mm.

Treatment with Alprostadil 0.05 micrograms/kg/min was instituted, 12 hours after the initiation of the therapy the infant showed marked psychomotor agitation and desaturation, which is why the Alprostadil administration is interrupted for 2 hours and re-introduced with half the initial dosage for 2 hours after which it is returned to the initial dose which is well tolerated.

Control echocardiography confirmed that the ductus arteriosus remained open and that the caliber of the descending aorta was slightly increased compared to the previous day.

Laboratory tests were within normal limits.

During the genetic consultation, a child with facial asymmetry, displaced ears, bulbous nose, a café au lait spot, an extended Mongolian spot, sacral fossa, ventricular septal defect, aortic arch observation was described. Analyzes are collected for MLPA microdeletions. Hypoplasia of the thymus from CT angiography raises the suspicion of microdeletion 22q11.2 – DiGeorge syndrome.

He is transferred to IRCCS Policlinic San Donato, Milan, Italy, with the serious general condition, afebrile, oxygen-dependent, fed by mixed gavage, with breast milk and powdered milk formula, systolic murmur grade IV/ 6 present on the entire precordial area with interscapular irradiation, HR = 130-170 b/min, SpO<sub>2</sub> (+) = 78 – 95% under mask oxygen therapy (flow = 2-3 l/min), BP 109 / 63 mmHg, temperature body = 36.9 degrees C, weight 4950 grams.

The treatment was maintained with Alprostadil 0.05 micro gr/kg/min, Spironolactone 6.25 mg/day and Furosemide 5 mg x 2/day.

## DISCUSSIONS

The interrupted aortic arch is considered to be the most severe form of coarctation of the aorta, be-

ing a congenital ductal-dependent cardiac malformation, which in the neonatal period, in the absence of rapid diagnosis and the initiation of appropriate therapy, presents a high mortality rate [3,17,18,19], likewise in our case, the delay or lack of diagnosis and initiation of drug therapy until the time of surgery would have led to the infant's death.

Multiple cardiac malformations are associated with interrupted aortic arch, most studies show association with ventricular septal defect, atrial septal defect, ductus arteriosus, aortopulmonary window, right ventricle with double orifice, aortic valve atresia, valvular or sub valvular aortic stenosis [4,20,21]. As well as in the case presented by us, the patient associated multiple complex congenital cardiac anomalies, ventricular septal defect of broad muscular type, the ascending aorta and the curve of the aorta had a hypoplastic and interrupted aspect, the descending aorta with apparent origin from the branch left pulmonary artery plus reduced caliber.

Ventricular septal defect is the most common congenital heart malformation, with a prevalence of 5.7 per 1000 live newborns [22,23,24], it is associated in a significant percentage (80%-90%) with the arch interrupted aortic [25,26], this major association was also identified in the case presented by us, where the patient associated a 5 mm wide muscular ventricular septal defect.

Hypoplasia of the thymus can be transient or permanent, a transient hypoplasia is found in the case of premature newborns, in malnutrition, stress, pregnancy, infections, radiation and a hypoplasia or aplasia of the thymus permanently in the case of genetic pathologies, the most frequent, hypoplasia thymus is associated with DiGeorge syndrome [27,28,29,30,31], during the genetic examination, due to the hypoplasia of the thymus shown in the angio-CT, the suspicion of DiGeorge syndrome was also raised in our patient, which is why they collected analyzes for genetic testing.

This duct-dependent congenital heart malformation is associated with multiple cardiac and genetic abnormalities, [3,9] a statement that is also proven

in the case presented by us, the patient being diagnosed with complete duct-dependent congenital heart malformation, interrupted aortic arch, persistence of ductus arteriosus, wide muscular ventricular septal defect, hypoplasia of the ascending aorta, observed with DiGeorge syndrome.

Keeping the arterial channel open in duct dependent congenital heart pathologies is an essential medical act, the administration of prostaglandin E1 maintains or reopens the arterial channel [33,34], and in the case presented by us, after performing the echocardiography, the first therapeutic measure was represented by administration of Alprostadil in a dose of 0.05 micrograms/kg/min.

Adverse effects of prostaglandin E1 administration are well known, their incidence varies according to studies from 21.5% to 33%, [35,36,37] among the frequent side effects, in our case, 12 hours after initiation of therapy with Alprostadil, psychomotor agitation and desaturation were reported, which is why the administration of Alprostadil was interrupted for 2 hours, then it was resumed at a dose of 0.025 micro grams/kg/min for 2 hours, later returning to the initial dose, the functionality of the arterial channel was confirmed by echocardiography.

## CONCLUSIONS

Interrupted aortic arch is a severe, duct-dependent congenital heart malformation with a rare incidence but high morbidity that associates with multiple other cardiac abnormalities and genetic syndromes. It requires a diagnosis as quickly as possible, preferably intranasally, so that the couple can decide together with the medical team the next steps towards a more effective therapeutic management. Postnatally, increased attention from the first minutes of life on the evolution of the child's general condition, together with a careful clinical examination, can help raise the suspicion of congenital heart malformation, at a less advanced stage, after which the child is sent to the cardiology clinic, where he will benefit from correct diagnosis and treatment.

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## REFERENCES

- Ramirez Alcantara J, Mendez MD. Interrupted Aortic Arch. SttPearlsPublishing, 2022. PMID: 30422497
- Varghese R, Saheed SB, Omoregbee B, Ninan B, Pavithran S, Kothandam S. Surgical Repair of Interrupted Aortic Arc and Interrupted Pulmonary Artery. *Ann Thorac Surg*. 2015 Dec;100(6):e139–40. doi:10.1016/j.annthoracsurg.2015.06.001. PMID:26652572
- Patel DM, Maldjian PD, Lovoulos C. Interrupted aortic arch with post-interrupted aneurysm and bicuspid aortic valve in an adult: a case report and literature review. *Radiol Case Rep*. 2015 Oct;10(3):5-8 doi:10.1016/j.radcr.2015.06.001
- Rodica Toganel. Abordarea diagnostica si terapeutica a afectiunilor cardiace congenitale la copil. Targu Mures: University Press, 2021
- Mishra PK. Management strategies for interrupted aortic arch with associated anomalies. *Eur J Cardiothorac Surg*. 2009;35(4):569-76. <https://doi.org/10.1016/j.ejcts.2008.12.044>
- Brown JW, Ruzmetov M, Okada Y, Vijav P, Rodefeld MD, Turrentine MW. Outcomes in patients with interrupted aortic arch and associated anomalies a 20 year experience. *Eur J Cardiothorac Surg*. 2006;29(5):666-73. <https://doi.org/10.1016/j.ejcts.2006.01.060>
- Steidele R.J. Sammig. Verchiedener in der chirurg 2. *Prakt Lehrschule Gemachten Beob.* 1778

8. McCrindle BW, Tchervenkov CI, Konstantinov IE, Williams WG, Neirrotti RA, Jacobs ML et al. Congenital Heart Surgeons Society. Risk factors associated with mortality and interventions in 472 neonates with interrupted aortic arch: a Congenital Heart Surgeons Society study. *J Thorac Cardiovasc Surg.* 2005;129(2):343-50. <https://doi.org/10.1016/j.jtcvs.2004.10.004> PMID:15678045
9. Konstantinov IE, Karamlou T, Williams WG, Quaegebeur JM, del Nido PJ, Spray TL, Caldarone CA. Congenital Heart Surgeons Society. Surgical management of aortopulmonary window associated with interrupted aortic arch: a Congenital Heart Surgeons Society study. *J Thorac Cardiovasc Surg.* 2006;131(5):1136-41. doi: 10.1016/j.jtcvs.2005.03.051 PMID :16678601
10. Luciani GB, Ackerman RJ, Chang AC, Welles WJ, Starnes VA. One – stage repair of interrupted aortic arch, ventricular septal defect, and subaortic obstruction in the neonate: a novel approach. *J Thorac Cardiovasc Surg.* 1996;111(2):348-58. doi:10.1016/s0022-5223(96)70444-0 PMID:8583808
11. Park MK, Salamat M, Park s Pediatric Cardiology For Practitioners, Seventh Edition. Philadelphia: Elsevier, 2020
12. Keane JF, Lock JE, Fyler DC. Nadas Pediatric Cardiology, Second Edition, Saunders. Philadelphia: Elsevier Inc, 2006
13. Marginean Claudiu, Malformatii cardiace fetale, Diagnostic si prognostic. Targu Mures: University Press, 2016
14. Volpe P, Marasini M, Caruso G, Gentile M. Prenatal diagnosis of interruption of the aortic arch and its association with deletion of chromosome 22q11. *Ultrasound Obstet Gynecol.* 2002;20(4):327-31. <https://doi.org/10.1046/j.1469-0705.2002.00818> PMID:12383312
15. Abuhamad AZ, Chaoui R. A practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts, second edition, Wolters Kluwer Health/ Lippincot, Williams&Wilkins, 2010.
16. Konstantinov IE, Karamlou T, Blackstone EH, Mosca RS, Lofland GK, Caldarone CA et al. Truncus arteriosus associated with interrupted aortic arch in 50 neonates: a Congenital Heart Surgeons Society study. *Ann Thorac Surg.* 2006;81(1):214-22. doi:10.1016/j.athoracsur.2005.06.072. PMID:16368368
17. Yasui H, Kado H, Yonenaga K, Kawasaki S, Shiokawa Y, Kouno H et al. Revised technique of cardiopulmonary bypass in one -stage repair of interrupted aortic arch complexes. *Ann Thorac Surg.* 1993;55(5):1166-71. [https://doi.org/10.1016/0003-4975\(93\)90026-e](https://doi.org/10.1016/0003-4975(93)90026-e). PMID:8494427
18. Serraf A, Lacour- Gayet F, Robotin M, Bruniaux J, Sousa – Uva M, Roussin R, Planche C. Repair of interrupted aortic arch – a ten – year experience. *J Thorac Cardiovasc Surg.* 1996;112(5):1150-60. doi:10.1016/S0022-5223(96)70128-9 PMID: 8911311
19. LaPar DJ, Baird CW. Surgical Considerations in interrupted Aortic Arch. *Semin Cardiothorac Vasc Anesth.* 2022;(3):278-84. doi:10.1177/1089253218776664 PMID:29774793
20. Messner G, Reul GJ, Flamm SD, Gregoric ID, Opfermann UT. Interrupted aortic arch in an adult single-stage extra-anatomic repair. *Tex Heart Inst J.* 2002;29(2):118-21. PMID:12075868
21. Barcin C, Kursaklioglu H. Interrupted aortic arch and aortic coarctation: tow similar entities. *Tex Heart Inst J.* 2010;37(1):134. PMID:20200652
22. Cox K, Algaze-Yojay C, Punn R, Silverman N. The Natural and Unnatural History of Ventricular Septal Defects Presenting in Infancy: An Echocardiography – Bas Review. *J Am Socof Echocardiog.* 2020;33(6):763-70. doi:10.1016/j.echo.2020.01.013. PMID:32249125
23. Saxena A, Mehta A, Sharma M , Salhan S, Kalaiyani M, Ramakrishnan S, Juneja R. Birth prevalence of congenital heart disease: a corss – sectional observational study from North India. *Ann Pediatr Cardiol.* 2016;9(3):205-9. doi:10.4103/0974-2069.189122 PMID:27625516
24. Penny DJ, Vick GW 3<sup>rd</sup>. Ventricular septal defect. *Lancet.* 2011;377(9771):1103-12. doi:10.1016/S0140-6736910)61339-6. PMID:21349577
25. Fridman WF, Silverman N. Congenital heart disease in infancy and childhood. In: Braunwald E, Zipes DO, Libby P, editors. Heart disease: a textbook of cardiovascular medicine. Vol. 2. Toronto: Saunders, Philadelphia, 2001
26. Goo HW, Park IS, Ko JK, Kim YH, Seo DM, Yun TJ et al. CT of congenital heart disease: normal anatomy and typical pathologic conditions. *Radiographics.* 2003;23:S147–65. doi: 10.1148/RG.23si035501. PMID: 14557509
27. Amatuni GS, Currier RJ, Church JA, Bishop T, Grimbacher E, Nguyen A et al. Newborn screening for severe combined immunodeficiency and T-cell Lymphopenia in California, 2010-2017. *Pediatrics.* 2019;143(2):e20182300. doi:10.1542/peds.2018-2300 PMID:30683812
28. Bhalla P, Wysocki CA, van Oers NSC. Molecular Insights Into the Causes of Human Thymic Hypoplasia With Animal Models. *Front Immunol.* 2020;11:830. doi:10.3389/fimmu.2020.00830. PMID: 32431714.
29. Kwan A, Abraham RS, Currier R, Brower A, Andruszewski K, Abbott JK et al. Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA.* 2014;312(7):729-38. doi: 10.1001/jama.2014.9132 PMID:25138334
30. Gruver AL, Sempowski GD. Cytokines leptin, and stress-induced thymic atrophy. *J Leukocyte Biol.* 2008;84(2):915-23. doi:10.1189/jlb.0108025.. PMID: 18495786
31. Chaudhry MS, Velardi E, Dudakov JA, Van Den Brink MRM. *Thymus: the next (re)generation. Immunol Rev.* 2016;271(1):56-71. doi: 10.1111/imr.12418. PMID: 27088907
32. Belkaya S, Silge RL, Hoover AR, Medeiros JJ, Eitson JL, Becker AM et al. Dynamic modulation of thymic microRNAs in response to stress. *PLoS ONE.* 2011;6(11):e27580. doi: 10.1371/journal.pone.0027580. PMID: 22110677
33. Hew MR, Gerriets V. Prostaglandin E1. StataPearls Publishing, 2022. PMID: 31536236
34. Roehl SL, Townsend RJ. Alprostadil (prostin VR Pediatric Sterile Solution, The Upjohn Company). *Drug Intell Clin Pharm.* 1982;16(11):823-32
35. Costabile RA, Mammen T, Hwang K. An overview and expert opinion on the use of alprostadil in the treatment of sexual dysfunction. *Expert Opin Pharmacother.* 2008;9(8):1421-9. doi: 10.1177/106002808201601103. PMID: 6756848
36. Hallidie – Smith KA. Prostaglandin E1 in suspected ductus dependent cardiac malformation. *Arch Dis Child.* 1984;59(11):1020-6. doi: 10.1136/ADC.59.11.1020. PMID:6542338
37. Lewis AB, Freed MD, Heymann MA, Roehl SK, Kensey RC. Side effects of therapy with Prostaglandin E1 in infants with critical congenital heart disease. *Circulation.* 1981;64(5):893-8. doi: 10.1161/01.CIR.64.5.893. PMID:7285304