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Truncus Arteriosus and Digeorge Syndrome: A Case Report and Literature Review

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Abstract **Case Report**

Truncus arteriosus is a rare congenital heart defect in which a single arterial trunk emerges from the heart, giving rise to the coronary arteries, pulmonary arteries, and the aorta. Its association with DiGeorge syndrome (also known as 22q11 microdeletion syndrome) is common. In addition to conotruncal cardiac malformations, this syndrome is associated with facial dysmorphia, thymic agenesis, absence of parathyroid glands with hypocalcemia. The diagnosis of truncus arteriosus should be clinically suspected in the presence of refractory cyanosis and a double cardiac murmur, and confirmed by echocardiography and/or thoracic CT angiography. A precise morphological description of the malformation and associated anomalies is mandatory to assist the surgeon in selecting the optimal timing and surgical technique. Thoracic CT angiography, as a complement to echocardiography, fulfills this objective. The prognosis without treatment is poor, with high mortality due to cardiac failure and severe pulmonary arterial hypertension. Treatment is surgical and consists essentially of closing the interventricular communication, detaching the pulmonary arteries from the truncus, and restoring continuity between the right ventricle and the pulmonary arterial branches, within the first three months of life. Prognosis has now improved thanks to new surgical techniques. We report the case of a three-month-old child admitted for afebrile seizures with hypocalcemia. Chest radiograph showed cardiomegaly, and echocardiography raised suspicion of truncus arteriosus. The diagnosis was confirmed by thoracic CT angiography with better characterization, revealing a Van Praagh type A2 truncus arteriosus with interventricular communication, right aortic arch, anomalies of the supra-aortic trunks with a retro-esophageal left subclavian artery. The diagnosis of 22q11 microdeletion syndrome was subsequently confirmed.

Keywords: Truncus arteriosus; Common arterial trunk; Conotruncal; Right aortic arch; 22q11 microdeletion; DiGeorge syndrome; Thoracic CT angiography.

INTRODUCTION

Common arterial trunk, also known as truncus arteriosus, is a rare cyanogenic congenital heart disease, representing 0.3 to 4% of congenital cardiac malformations. It consists of a single arterial trunk emerging from the ventricles, giving rise to the coronary arteries, the aorta, and one or two pulmonary arteries. [1-6]

It is caused by the absence of conotruncal septation during embryonic development. This is a serious condition with high mortality, notably due to cardiac failure and severe pulmonary arterial hypertension, which become irreversible early on. [1-6]

On the other hand, 22q11.2 deletion syndrome, formerly known as DiGeorge syndrome, is also a rare congenital chromosomal disorder. The phenotype varies widely and notably includes characteristic facial dysmorphia, thymic agenesis, absence of parathyroid glands with hypocalcemia, conotruncal cardiac malformations, cleft palate, and immune deficiency, among others. The association of truncus arteriosus with 22q11 microdeletion syndrome is common (35% to 40% of patients diagnosed with TA). [6-11]

CLINICAL CASE

This is a 3-month-old child admitted for afebrile seizures complicated by status epilepticus. The medical history confirmed a probable case of cardiac malformation in a cousin who died at a young age.

Physical examination noted retrognathism and low-set ears (Figure 1). The clinical course was marked by the onset of respiratory distress. Chest radiograph showed vascular congestion, an ovoid cardiac silhouette, and a narrow superior mediastinum (Figure 2). Laboratory workup revealed refractory hypocalcemia with hypoparathyroidism, and normochromic normocytic anemia. Echocardiography was requested and raised suspicion of truncus arteriosus type III according to the Collett and Edwards classification. Thoracic CT angiography was then requested for confirmation and better characterization. It revealed: a single arterial trunk straddling both ventricles, giving rise to the aorta which continues its course, and to the right and left pulmonary arteries which arise separately in a contiguous manner from its postero-lateral left aspect (truncus arteriosus type II of Collett and Edwards and A2 of Van Praagh), with an interventricular communication measuring 11 mm. There was also a right aortic arch giving rise to the supra-aortic trunks each separately (absence of the brachiocephalic arterial trunk), in the following order (from proximal to distal): left common carotid artery, right common carotid artery, right subclavian artery, and then the left subclavian artery. The latter has a retro-esophageal course with upstream dilatation of the esophagus (left Arteria Lusoria). The common carotids thus arise separately from the aortic arch anterior to the subclavian arteries. There was also mild cardiomegaly, dilated appearance of the pulmonary arteries and pulmonary veins (especially on the left) with the presence of a common collector, and a small inter-atrial communication. Thymic hypotrophy was also noted. On the parenchymal window, consolidation foci were noted at the bilateral basal and hilar levels and at the apex of the left upper lobe (Figures 3 and 4). During hospitalization, the child contracted a nosocomial infection with Enterococcus faecium which responded well to antibiotic therapy. Surgical management was planned for the following month.

Fig. 1: Low-set ears with retrognathism

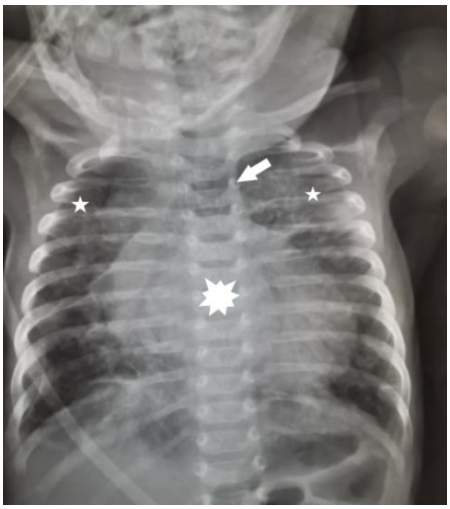

Fig. 2: Chest radiograph: Cardiomegaly with an "egg-on-its-side" appearance (multipointed star), vascular congestion predominating in the parahilar regions and bilateral upper lobes (stars). Thymic hypoplasia with a narrow superior mediastinum (arrow).

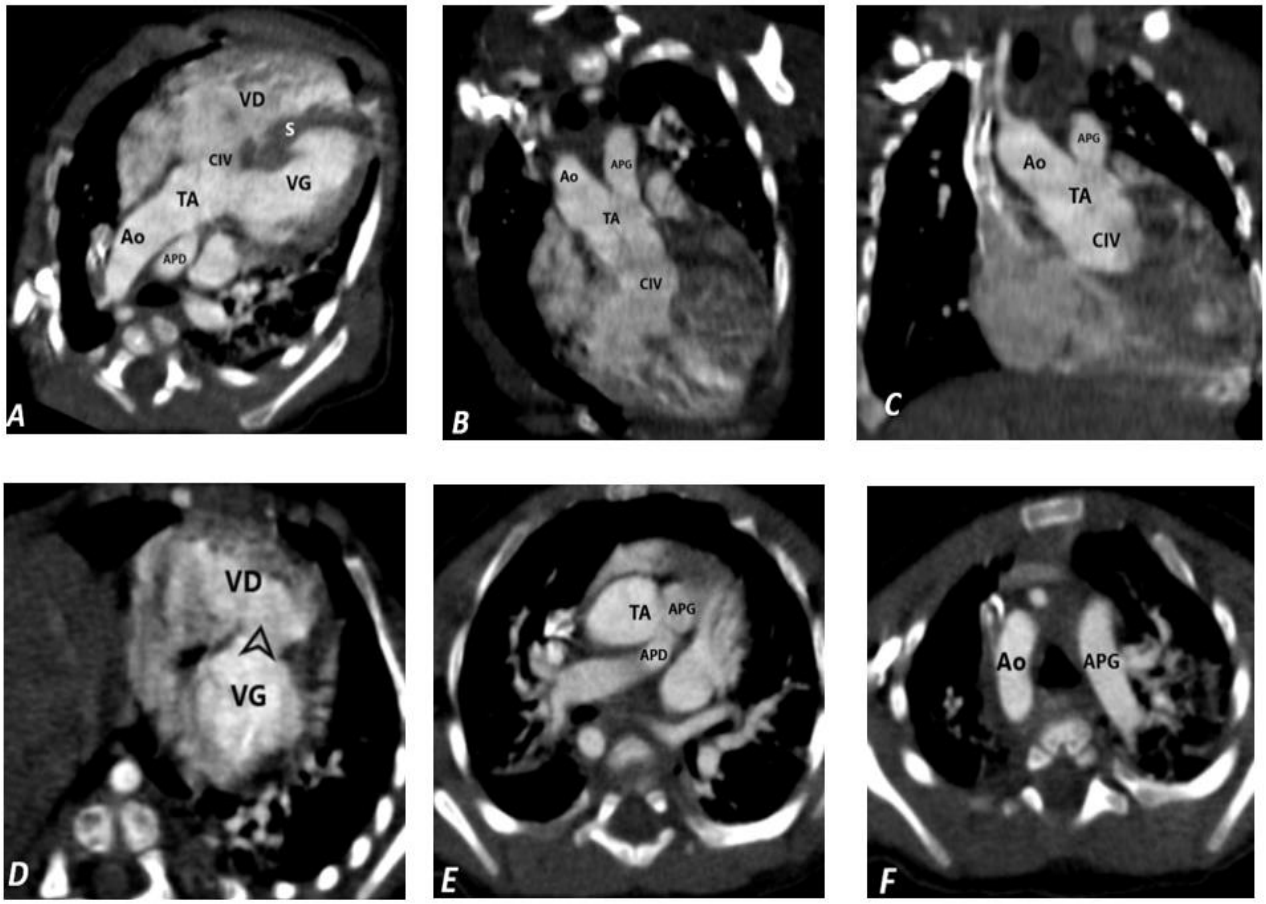

Fig. 3: Thoracic CT angiography in the arterial phase: Truncus arteriosus straddling both ventricles above a VSD, giving rise to the aorta and the two pulmonary arteries. A: Oblique axial section nearly parallel to the four-chamber plane; B: Oblique coronal section; C-D: Oblique coronal sections through the interventricular septum plane (C) and axial section close to the short-axis plane (D): VSD (arrow) with anterosuperior location immediately below the truncal valve. E: Axial section: pulmonary arteries emerging from the truncus arteriosus. F: Right aortic arch

TA: Truncus arteriosus; Ao: Aorta; APD: Right pulmonary artery; APG: Left pulmonary artery; CIV: Ventricular septal defect; VD: Right ventricle; VG: Left ventricle; S: Interventricular septum

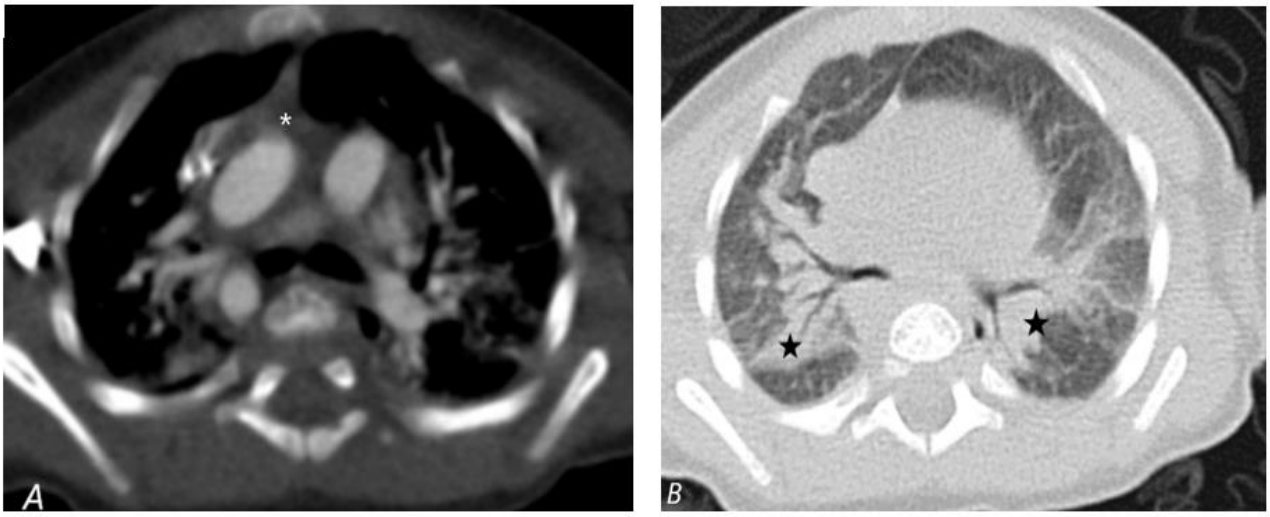

Fig. 4: Thoracic CT angiography in mediastinal window

A: thymic hypoplasia (*) and parenchymal window B: pulmonary parenchymal consolidation foci

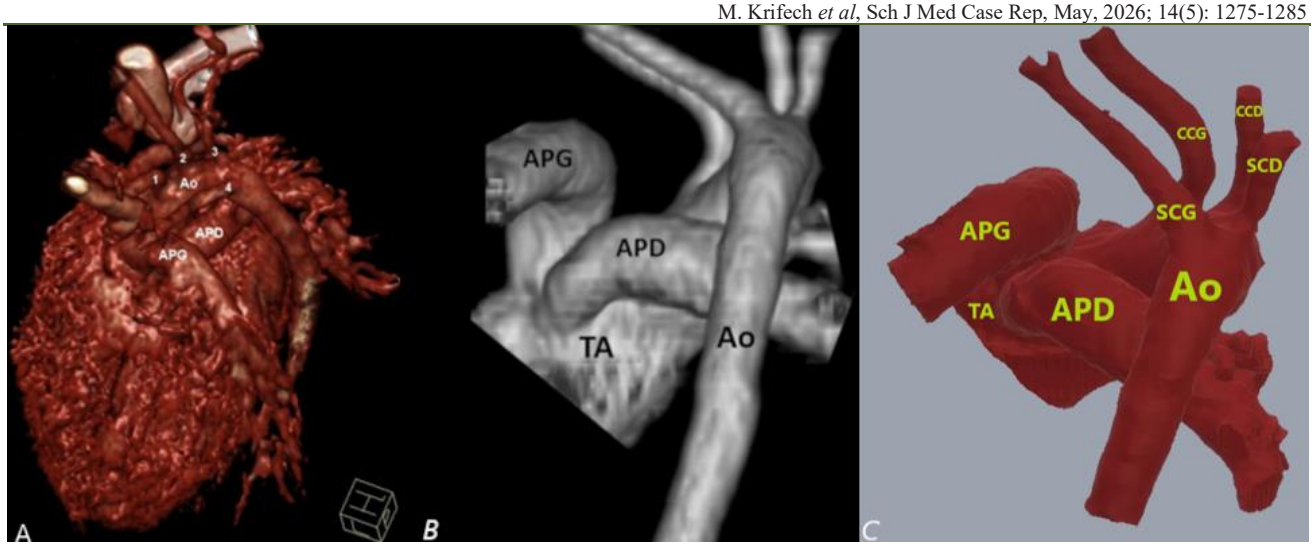

Fig. 5: 3D reconstructions:
A: Left postero-superior view; B: Posterior view; C: Postero-superior view.

1: Left common carotid artery (CCG); 2: Right common carotid artery (CCD); 3: Right subclavian artery (SCD); 4: Left subclavian artery (SCG);

TA: Truncus arteriosus; Ao: Aorta; APD: Right pulmonary artery; APG: Left pulmonary artery

DISCUSSION

Definition

Common arterial trunk, or truncus arteriosus, is a rare cyanogenic congenital heart disease defined by the emergence of a single large vessel from the base of the heart (instead of two) above a single semilunar valve called the truncal valve, and it almost always arises above a large interventricular communication (VSD). This trunk gives rise to the coronary arteries, the aorta, and one or two pulmonary arteries (Figure). [12]

Three malformations are excluded from this definition:

- The presence of a main pulmonary arterial trunk without branching, with two pulmonary arteries emerging from the ascending aorta, descending aorta, or supra-aortic trunks: this anomaly is considered a type of pulmonary atresia with VSD
- One branch of the pulmonary artery arises from the ascending aorta and the other from the main pulmonary arterial trunk: this malformation was formerly called hemitruncus arteriosus, although it has no known direct embryological or morphological relationship with the common arterial trunk
- Aortopulmonary windows.

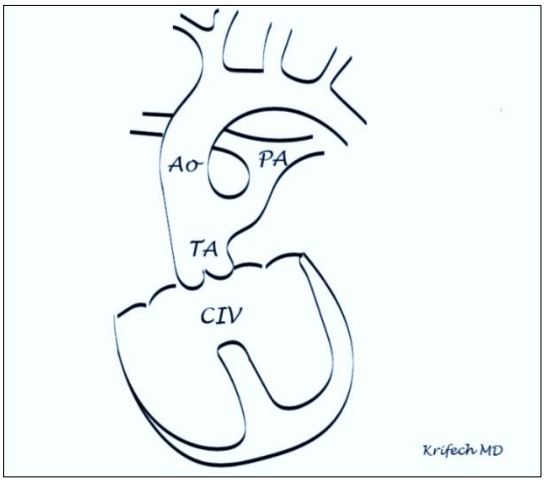

Figure 6: Simplified diagram of a Truncus Arteriosus

TA: Truncus Arteriosus; Ao: Aorta; PA: Pulmonary artery

Epidemiology

Truncus arteriosus is a rare malformation with an incidence ranging from 4 to 11 per 100,000 live births [4,16-18]. It accounts for 0.3 to 4% of congenital heart defects [4-6]. It is a serious pathology, rapidly causing cardiac failure and severe pulmonary arterial hypertension, which become irreversible early on. [4]

DiGeorge syndrome or 22q11.2 microdeletion has an estimated prevalence of 1 in 4,000 to 6,000 live births. The association of DiGeorge syndrome or 22q11.2 microdeletion with truncus arteriosus is common (35 to 40% in patients diagnosed with TA). [6-11]

Maternal diabetes has been identified as an independent risk factor for the development of TA. [6] In the recent series by Gerardo *et al* [20]: among 98 infants presenting with at least one non-syndromic congenital heart defect, eight (8.2%) carried the 22q11.2 deletion and all had a conotruncal-classified heart defect, including two truncus arteriosus cases.

Furthermore, in the series by Minier *et al* [10], truncus arteriosus was found, among other conotruncal malformations, in 2 out of 52 patients (3.8%) diagnosed with 22q11.2 microdeletion by the FISH method. These data confirm the importance of systematically screening for the 22q11.2 deletion in every newborn with a congenital heart defect, particularly of the conotruncal subtype, even in the absence of other manifestations. [20]

However, the true prevalence of congenital heart disease in 22q11.2 deletion syndrome is unknown and varies according to the population studied (75 to 80% in older studies, 48.5% to 79% in more recent studies). [11]

Embryology and Genetics

TA results from the absence of septation of the embryological arterial trunk, which normally occurs at the end of the 5th week, and from the absence of the proximal conal septum. This is why the term 'persistent truncus arteriosus' was previously used.

Normal partitioning of the cardiac bulb and arterial trunk is initiated by two bulbar ridges and two aorticopulmonary ridges that are continuous with each other. They fuse along a spiral course forming the spiral and aorticopulmonary septa, which divide the bulb and arterial trunk into the ascending aorta and pulmonary trunk. [23]

Truncus arteriosus results from abnormal development of neural crest cells, which are responsible for this septation [4][24], with involvement of the Tbx1 gene. [25]

Some authors refer to the concept of aortic or pulmonary dominance of TA based on embryological data and argue that using the term 'conotruncal' anomaly alone is insufficient to understand the development of the various lesions. [26-27]

Anatomy and Classifications

The common arterial trunk is large, and the aortic arch that follows it curves to the right in 25% of cases, with a mirror image of the supra-aortic trunks. Other aortic arch malformations may be associated: double arch, or right arch with retro-esophageal left subclavian artery. The horizontal aorta may be frankly hypoplastic or completely atretic with a patent ductus arteriosus.

This common arterial trunk has a so-called truncal valve, which may be tricuspid, quadricuspid, or bicuspid (in 69%, 21%, and 9% of cases, respectively) [11]. It is very frequently dysplastic (insufficiency in nearly 50% of cases and/or stenosis in nearly 20% of cases). [12]

This valve straddles an interventricular communication that is almost always present [4-5], usually very wide, generally anterosuperior, high just below the valve, and non-restrictive. Sometimes the arterial annulus is displaced to the right with hypoplasia of the ventricle contralateral to the common trunk [9]. The reverse situation, or levoposition, is rarer. [10]

Anomalies of origin and distribution of the coronary arteries are, by contrast, very frequent and must be mentioned before surgical repair. [4,6]

The origin of the pulmonary arteries is variable and forms the basis of numerous anatomical classifications of TA, the most classic being that of Collett and Edwards [2,4,6]:

- Type 1: A single pulmonary trunk and an ascending aorta both arising from the TA.
- Type 2: The right and left pulmonary arteries emerge together, close to each other, from the dorsal aspect of the TA.
- Type 3: One or both pulmonary arteries emerge independently from one side of the TA.
- Type 4: There are no pulmonary arteries. Collateral arteries from the descending aorta supply the lungs (also called pseudotruncus arteriosus, not to be confused with the former hemitruncus arteriosus). This type is better described as pulmonary atresia with a ventricular septal defect. [2,4]

Currently the most widely used classification is that of Van Praagh *et al*., which distinguishes group A with VSD from the exceptional group B forms with an intact interventricular septum. [1,4-6]:

- Type A1: An individualized pulmonary trunk is present from which the right and left pulmonary arteries arise; corresponds to Collett and

Edwards type 1. This is the most common type (60 to 70% of cases).

- Type A2: The two pulmonary arteries arise separately from the common trunk, generally from its posterior aspect; corresponds to Collett and Edwards types 2 and 3; this type accounts for 20 to 30% of cases.
- Type A3 is very rare. The common trunk gives rise to only one branch, generally the right; vascularization of the other lung is provided by one or more arteries arising from the arch or descending thoracic aorta, or even from the ductus arteriosus or aortopulmonary collaterals.
- Type A4 is type A1 with interruption of the aortic arch or coarctation and a large ductus arteriosus.

Anderson further subdivided TA into two categories: forms with aortic dominance versus forms with pulmonary dominance. In the aortic-dominant form, the pulmonary artery branches often arise separately but adjacent from the truncal root. In the pulmonary-dominant form, there is often an aortic interruption or coarctation, and the pulmonary arteries often arise from each side of the truncal root, with a persistent ductus arteriosus often present. [6] This simplified approach based on the concept of aortic or pulmonary dominance of the intrapericardial component of the common trunk was discussed by several authors including Praagh and Calder, then promoted by Russell *et al* [6], describing it as simple and highly useful. [6,27] According to the latter, the observation of equal-sized aortic and pulmonary components is rarely reported and justifies consideration of a third category, namely the balanced common arterial trunk. [27]

Associated Anomalies

Several anomalies are commonly associated with TA, including severe aortic coarctation, interruption of the aortic arch (20% of cases) (notably type B, generally after the origin of the left common carotid artery), right aortic arch with a mirror image of the supra-aortic trunks (30%), or retro-esophageal left subclavian artery, and more rarely a double aortic arch.

Persistence of the ductus arteriosus is an integral part of the circulation in cases of pulmonary dominance and may exist in less than 10% of cases outside arch interruption.

An inter-atrial communication, anomalies of pulmonary or systemic venous return, single ventricle, atrioventricular canal, may also be found. [4-6]

Clinical Presentation and Association with DiGeorge Syndrome

Early on, often before 1 month of age, signs of left-to-right shunt may reveal TA: notably dyspnea and feeding fatigue, soon accompanied by other signs of cardiac failure: sweating, hepatomegaly, hypotrophy.

Although TA is a cyanogenic congenital heart disease, cyanosis may be absent especially in the first days of life. It may also be mild and go unnoticed. More significant central cyanosis is observed in children with persistently elevated pulmonary vascular resistance or stenosis of the pulmonary arteries. Heart sounds are intense and there is a constant left parasternal systolic murmur. The findings of physical examination reflect the state of congestive cardiac failure, which depends on the magnitude of pulmonary flow and the presence or absence of truncal valve insufficiency. Electrocardiographic data are not specific. The clinical course rapidly progresses toward cardiac failure and severe pulmonary arterial hypertension, which become irreversible early on. [4,6]

On the other hand, 22q11.2 deletion syndrome is associated with thymic and parathyroid hypoplasia, conotruncal-type congenital heart disease, and characteristic facial dysmorphia. This syndrome is characterized by great clinical heterogeneity with inter- and even intra-familial variability. Among the most common clinical features: facial dysmorphia, congenital heart disease, hypocalcemia, renal anomalies, skeletal anomalies, psychomotor delay, cleft palate, behavioral/psychiatric disorders, and immune deficiency. [10,11]

The cardiac anomalies most commonly observed in 22q11.2 deletion syndrome include: Cono truncal heart diseases (notably: tetralogy of Fallot, truncus arteriosus, type B interruption of the aortic arch), ventricular and atrial septal defects. [11,29,30] In this regard, Bonnet *et al.,*[31] showed that 50% of Cono truncal heart diseases are related to DiGeorge syndrome. For truncus arteriosus, Bassil-Eter [4] mentioned that Van Praagh type A4 is particularly common in this syndrome. However, according to Peyvandi *et al*, a 22q11.2 deletion has been observed in all subtypes of truncus arteriosus. [32]

Aortic arch anomalies, whether isolated or not, are also very common (cervical aortic arch, double aortic arch, right aortic arch, and abnormal origin of the subclavian arteries), and according to Peyvandi *et al* [32], up to 68% of patients with a 22q11.2 deletion and an intracardiac anomaly have a simultaneous aortic arch anomaly. Aortic root dilatation has also been mentioned as well as other cardiac malformations. It should be noted according to various studies that 22q11 deletion status appears to be an independent risk factor for severity and mortality in congenital heart diseases, whether in truncus arteriosus or others, and the underlying mechanisms are not well understood at present [11]. Therefore, screening for this syndrome is recommended in the presence of any conotruncal heart disease. [10,11]

Facial dysmorphia is almost constant but may go unnoticed as it is most often subtle or even absent. It

includes hypertelorism, prominent nasal root, malar flattening, and small low-set ears. [10]

Severe immune deficiencies of DiGeorge syndrome are rare. Most often, there is a mild to moderate T-cell immunity deficiency. [33] Hypocalcemia is related to hypoparathyroidism (HPT), which may be total or partial, itself linked to parathyroid hypoplasia. It is not constant and its exact prevalence in individuals with 22q11.2 microdeletion is difficult to estimate. [33]

Imaging

The main goal of imaging is to make an early diagnosis of truncus arteriosus, with a precise description of its morphological characteristics, associated anomalies and factors that may hinder surgical intervention, and to assess possible complications in order to assist surgical planning. Imaging also plays a role in postoperative follow-up.

Chest Radiograph

Typically, chest radiograph demonstrates cardiomegaly and accentuation of pulmonary vascularization. Cardiomegaly is almost constant with an oval cardiac configuration but non-specific (sometimes described as an 'egg-on-its-side' appearance, also found in transposition of the great arteries and pulmonary atresia with intact septum, or a 'boot-shaped heart' more suggestive of tetralogy of Fallot). The middle arc is concave as in tetralogy of Fallot, except in Van Praagh type A1. Accentuation of pulmonary vascularization may be absent in forms with small pulmonary branches or in obstructive pulmonary vascular disease, and may be asymmetric. Atelectasis, emphysema, and pneumonia foci may be associated [3]. The abnormally high origin of the pulmonary arteries, notably the left pulmonary artery, may produce the 'hilar comma sign' (left pulmonary artery curving upward to form a left hilar comma) [4,34]. A narrow superior mediastinum may suggest Van Praagh type A2, and thymic absence or hypoplasia along with a right aortic arch should raise suspicion of DiGeorge syndrome. [5]

Thoracic CT Angiography

Thoracic CT angiography has the main advantages of accessibility, speed, and multiplanar analysis with 3D volumetric reconstructions, offering a better anatomical approach to complex congenital heart diseases.

Preoperatively, it allows confirmation of a true truncus arteriosus with its cardinal defining elements: the presence of a single large vessel arising from the base of the heart giving rise to the aortic arch and to at least one pulmonary artery. It also allows classification of the TA with a precise description of its branching divisions, the pulmonary arteries and their course, the state of the aortic arch (searching for interruption or coarctation) and its position, searching for MAPCAs (major aorto-pulmonary collateral arteries) (particularly in type A3), searching for a persistent ductus arteriosus, studying the disposition of the supra-aortic trunks, and studying the truncal valve and VSD.

The search for possible associated anomalies is an integral part of CT angiography objectives. Thus, description of the coronary arteries and their frequent anomalies is valuable for the surgeon. These may include: a high origin which may interfere with surgery (surgically dangerous proximity to the origin of the pulmonary branches, hence the interest for some authors in measuring the distance between this origin and the origin of the left main coronary artery), the presence of large collaterals crossing the right ventricular outflow tract anteriorly (to be specified before surgery), a single coronary artery, a retro-aortic course of the left coronary artery, an intramural course, or very rarely, an anomalous origin of the right coronary artery from the pulmonary artery. [36] Associated anomalies of the aortic arch may include: right aortic arch (30-33%) [5], interruption of the aortic arch (9-11%), or more rarely, double aortic arch [37]. Sometimes, a retro-esophageal left subclavian artery may be present. An inter-atrial communication, anomalies of pulmonary or systemic venous return, single ventricle, and atrioventricular canal may also be found. [35]

Morphological analysis by CT angiography can also infer the type of aortic or pulmonary predominance of the truncus arteriosus, and allows systematic evaluation of the state of the cardiac chambers and their walls, as well as the presence of signs of pulmonary arterial hypertension.

Postoperatively, CT angiography allows assessment of the state of the pulmonary homograft and the neo-aortic valve. In addition, the aortic arch and pulmonary arteries should be evaluated for any residual or recurrent stenosis. [18]

For the classification of truncus arteriosus, the Van Praagh classification is widely used, and recently the 'Congenital Heart Surgery Nomenclature and Database Project' proposed the use of a modified Van Praagh classification with a surgical perspective. This classification divides truncus arteriosus into three categories [39]:

- Type 1, with confluent or nearly confluent pulmonary arteries, also named 'large Aorta type' (Van Praagh type A1, A2 and Collett and Edwards type I, II, III)
- Type 2, with absence of one pulmonary artery or the 'large Aorta type with absent pulmonary artery' (Van Praagh type A3)
- Type 3, with interrupted aortic arch or severe coarctation representing the 'large Pulmonary Artery type' (Van Praagh type A4).

Regarding DiGeorge syndrome, as mentioned in the anatomy section, certain anomalies may point toward the diagnosis: absence or hypoplasia of thymic tissue, aortic arch anomalies (cervical aortic arch, double aortic arch, right aortic arch, type B aortic arch interruption, and abnormal origin of the subclavian arteries [aberrant, retro-esophageal left subclavian artery, isolated origin of right subclavian artery). [18]

Magnetic Resonance Imaging (MRI)

Magnetic resonance imaging (MRI) is an important alternative and non-irradiating imaging modality. It allows study of the arterial axes with description of the pulmonary arterial branches. It is the modality of choice when kinetic or flow imaging is needed. However, increased imaging time with sometimes the need for general anesthesia. [40]

Echocardiography

This is the first-line modality for the evaluation of TA as for all congenital heart diseases, but it is operator-dependent and does not allow adequate study of extracardiac structures. [5] It usually allows, after affirming the diagnosis of TAC, its complete morphological study and that of associated lesions. [25] In addition it allows its functional study. It must study the VSD, the truncal valve and its relationship to the atrioventricular valves, the type of truncus, and the origin of the coronary arteries with their distribution. [4]

Fetal Ultrasonography

The diagnosis of truncus arteriosus should not be missed on screening fetal ultrasound. However, it is not uncommon for it to be confused with tetralogy of Fallot with pulmonary atresia and major aortopulmonary collaterals. [6] The 'three-vessel view' is useful for diagnosis. [42] This is an orthogonal transverse section of the upper fetal mediastinum, where the oblique section of the pulmonary trunk, the transverse section of the ascending aorta, and the superior vena cava are arranged on a straight line with decreasing size. It is obtained by moving the probe cranially from the 'four-chamber view'. The diagnosis of TA is suspected when only two vessels are identified on this view. [4] The most common indication for prenatal screening is a family history of congenital heart disease, particularly conotruncal, or a family history of DiGeorge syndrome. Prenatal diagnosis allows prenatal counseling for parents and prepares for subsequent management by avoiding late diagnosis and potential complications (ischemic myocardial dysfunction, significant pulmonary arterial hypertension). [6] When the diagnosis of truncus arteriosus is made on fetal ultrasound, karyotyping is requested using the FISH method (fluorescence in situ hybridization) to confirm the 22q11 syndrome. [6]

Differential Diagnoses

- Aortopulmonary septal defect (APSD) or aortopulmonary window: a rare class of congenital cardiac malformations with a direct communication between the ascending aorta and the pulmonary trunk in their intrapericardial part in the presence of two individual semilunar valves.
- Main pulmonary arterial trunk without branching, with two pulmonary arteries emerging from the ascending aorta, descending aorta, or supra-aortic trunks: this anomaly is considered a type of pulmonary atresia with VSD. Cases where both pulmonary arteries have a single origin from the ascending aorta (SOPA: Single Origin of Pulmonary Arteries) can be observed. In these situations, the diagnosis may be confused with Van Praagh type A2, but it is important to note that in SOPA, two separate aortic and pulmonary valves are present.
- Former hemitruncus arteriosus: One branch of the pulmonary artery arises from the ascending aorta and the other from the main pulmonary arterial trunk in the presence of two normal semilunar valves.
- Tetralogy of Fallot: the most common cyanogenic congenital heart disease, defined by obstruction of the pulmonary outflow tract, an interventricular communication, an overriding aorta, and right ventricular hypertrophy.
- Pulmonary atresia with VSD: there is a blind pulmonary infundibulum and no pulmonary artery is seen arising from the great vessel.
- Origin of the right (or exceptionally left) pulmonary artery from the aorta: especially for types A3
- Isolated interruption of the aortic arch: especially for type A4 forms

Prognosis and Treatment Principles

The prognosis of truncus arteriosus without surgical intervention is very poor, with one-year mortality exceeding 80%. Adult survivors are extremely rare. Medical treatment is mainly symptomatic (including diuretics, ACE inhibitors, nitric oxide, inotropic support).

In common forms of truncus arteriosus where the ductus arteriosus is absent, the use of prostaglandin E2 is not necessary. In contrast, in rare pulmonary-dominant forms with interruption or coarctation of the aorta, a patent ductus arteriosus is often present. In these cases, prostaglandin E1 infusion should be initiated immediately after birth and continued until surgery.

Surgical treatment is the only effective treatment and includes closure of the interventricular communication (VSD), separation of the pulmonary arteries from the truncal root, closure of the resulting defect, restoration of continuity between the right ventricle and pulmonary arteries using a cryopreserved homograft or a valved heterograft conduit, replacement

of the truncal valve, and treatment of associated lesions. [47,48]

Several studies have emphasized that the association with DiGeorge syndrome is an independent poor prognostic factor. [11] Lifelong cardiac follow-up is necessary.

Comments on the Clinical Case

In our case, facial dysmorphia was unnoticed at the patient's admission as it was not particularly evident. The etiological workup for seizures revealed refractory hypocalcemia with hypoparathyroidism, which may point toward DiGeorge syndrome. Then, in the context of cardiomegaly and onset of respiratory distress, echocardiography identified truncus arteriosus. This justified complementary CT angiography for confirmation and better characterization, and it was only at this stage that the diagnosis of 22q11 microdeletion syndrome associated with truncus arteriosus was highlighted, particularly in view of the evident thymic hypoplasia, mild facial dysmorphia, and associated anomalies of the conotruncal malformation, notably: the right aortic arch with anomalous origin of the supra-aortic trunks. This allowed correction of the clinical management. In addition, CT angiography allowed reclassification of the truncus arteriosus and better characterization of the associated lesions. The location of the VSD was typical in the anterosuperior position, and the ASD (atrial septal defect) was minimal. There were no MAPCAs, and no anomaly of coronary artery origin was found, but the surgeon was informed that the truncus arteriosus was relatively short with emergence of both pulmonary arteries fairly close to it (which may constitute a surgical hindrance). Regarding supra-aortic trunk anomalies, these are described in the literature with possible left Arteria Lusoria in this association but with a higher frequency of mirror-image arrangement (with the presence of a left brachiocephalic arterial trunk). However, no coarctation or aortic narrowing was found. Regarding predominance, since there was no aortic interruption, one cannot speak of true pulmonary predominance despite the increased diameter of the pulmonary arteries; a balanced form may therefore be evoked. Regarding pulmonary consolidations, given their topography with non-systemic involvement and the presence of peribroncho-vascular thickening, a hemodynamic origin is primarily suggested, although an infectious origin cannot be excluded.

Conclusion

The association of 22q11 deletion syndrome with truncus arteriosus represents a true diagnostic and therapeutic challenge. Echocardiography remains the preferred initial modality for diagnosis; however, CT angiography and MRI allow better morphological assessment. Differential diagnosis can be difficult with other cardiovascular malformations, and the numerous anatomical variants require a thorough understanding of the proposed classifications, hence the importance of cross-sectional imaging in the diagnostic approach. It enables early diagnosis, avoidance of complications, and better planning of surgical treatment in order to improve prognosis for these patients, given that the 22q11 microdeletion is an independent severity factor in this association.

References

1. Van Praagh R, Van Praagh S. The anatomy of common aorticopulmonary trunk (truncus arteriosus communis) and its embryologic implications. A study of 57 necropsy cases. Am J Cardiol. 1965; 16:406-425.
2. Collett RW, Edwards JE. Persistent truncus arteriosus; a classification according to anatomic types. Surg Clin North Am. 1949;29:1245-1270.
3. Calder L, Van Praagh R, Van Praagh S, *et al.,*Truncus arteriosus communis. Clinical, angiocardiographic, and pathologic findings in 100 patients. Am Heart J. 1976; 92:23-38.
4. Bassil-Eter R, Acar P, Leonbon B. Common arterial trunk. EMC - Cardiol. 2007;2(4):1-9. doi:10.1016/S1166-4568(07)32772-1
5. Sharma A, Priya S, Jagia P. Persistent truncus arteriosus on dual source CT. Jpn J Radiol. 2016;34(7):486-493. doi:10.1007/s11604-016-0559-x
6. Chikkabyrappa S, Mahadevaiah G, Buddhe S, Alsaied T, Tretter J. Common Arterial Trunk: Physiology, Imaging, and Management. Semin Cardiothorac Vasc Anesth. 2019;23(2):225-236. doi:10.1177/1089253218821382
7. Goldmuntz E, Clark BJ, Mitchell LE, *et al.,* Frequency of 22q11 deletions in patients with conotruncal defects. J Am Coll Cardiol. 1998; 32:492-498.
8. McElhinney DB, Driscoll DA, Emanuel BS, Goldmuntz E. Chromosome 22q11 deletion in patients with truncus arteriosus. Pediatr Cardiol. 2003;24:569-573.
9. Iserin L, de Lonlay P, Viot G, *et al.,* Prevalence of the microdeletion 22q11 in newborn infants with congenital conotruncal cardiac anomalies. Eur J Pediatr. 1998; 157:881-884.
10. Minier F, Carles D, Pelluard F, Alberti EM, Stern L, Saura R. DiGeorge syndrome, retrospective study of 52 cases. Arch Pediatr. 2005;12(3):254-257. doi: 10.1016/j.arcped.2004.10.015
11. Goldmuntz E. 22q11.2 deletion syndrome and congenital heart disease. Am J Med Genet C Semin Med Genet. 2020;184(1):64-72. doi:10.1002/ajmg.c.31774
12. Kastler B *et al.,* Malformations of the pulmonary arteries. Cyanogenic heart diseases: MRI aspects. 32-015-F-70 EMC 2002
13. Viczaino A, Campbell J, Litovsky S, Van Praagh R. Single origin of right and left pulmonary artery branches from ascending aorta with nonbranching

main pulmonary artery: relevance to a new understanding of truncus arteriosus. Pediatr Cardiol 2002; 23:230-4.
14. Beitzke A, Shinebourne EA. Single origin of right and left pulmonary arteries from ascending aorta, with main pulmonary artery from right ventricle. Br Heart J 1980; 43:363-5.
15. Laura G, David W B. Truncus arteriosus and hemitruncus. Nadas' Pediatric Cardiology (Third Edition) 2025, Pages 478-484, ISBN 978-1-4557-0599-3, 2013
16. Freedom RM. Anomalies of aortopulmonary septation: persistent truncus arteriosus, aortopulmonary septal defect and Hemitruncus arteriosus. In: Freedom RM, Benson LN, Smallhorn JF, editors. Neonatal heart disease. London: Springer Verlag; 1992. p. 425-52.
17. Hoffman JI, Kaplan S. The incidence of congenital heart disease. J Am Coll Cardiol. 2002; 39:1890-1900.
18. Kumar P, Bhatia M. Role of CT in the Pre- and Postoperative Assessment of Conotruncal Anomalies. Radiol Cardiothoracic Imaging. 2022;4(3): e210089. doi:10.1148/ryct.210089
19. Keane JF, Fyler DC. Truncus arteriosus. In: Keane JF, Lock JE, Fyler DC, editors. NADAS' Pediatric cardiology. Philadelphia: WB Saunders-Elsevier; 2006. p. 767-71.
20. Gerardo E *et al.,* Frequency of chromosome 22q11.2 deletion among newborns with non-syndromic congenital heart defects from western Mexico. Bol Med Hosp Infant Mex. 2022;79(6) DOI: 10.24875/BMHIM.22000078
21. Lorenzo DB and col. A population-based study of the 22q11.2 deletion: phenotype, incidence, and contribution to major birth defects in the population. Pediatrics. 2003 Jul;112(1 Pt 1):101-7. doi: 10.1542/peds.112.1.101.
22. Oskarsdottir S, Vujic M, Fasth A. Incidence and prevalence of the 22q11 deletion syndrome: a population-based study in Western Sweden. Archives of Disease in Childhood. 2004;89(2):148-151. https://doi.org/10.1136/adc.2003.026880
23. Kamina P. Clinical Anatomy - Volume 3, Thorax, abdomen 4th edition. Maloine
24. Kirby ML, Gale TF, Stewart DE. Neural crest cells contribute to normal aorticopulmonary septation. Science 1983; 220:1059-61.
25. Lindsay EA, Vitelli F, Su H, Morishima M, Huynh T, Pramparo T, *et al.,* Tbx1 haplo-insufficiency in the DiGeorge syndrome region causes aortic arch defects in mice. Nature 2001; 410:97-101.
26. Anderson RH, Chaudhry B, Mohun TJ, *et al.,* Normal and abnormal development of the intrapericardial arterial trunks in humans and mice. Cardiovasc Res. 2012; 95:108-115.
27. Anderson RH, Mori S, Spicer DE, Brown NA, Mohun TJ. Development and morphology of the ventricular outflow tracts
28. Russell HM, Jacobs ML, Anderson RH, *et al.,* A simplified categorization for common arterial trunk. J Thorac Cardiovasc Surg. 2011; 141:645-653.
29. Campbell IM, Sheppard SE, Crowley TB, McGinn DE, Bailey A, McGinn MJ, McDonald-McGinn DM. What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. American Journal of Medical Genetics. Part A. 2018;176(10):2058-2069. https://doi.org/10.1002/ajmg.a.40637
30. Cancrini C, Puliafito P, Digilio MC, Soresina A, Martino S, Rondelli R, *et al.,* Clinical features and follow-up in patients with 22q11.2 deletion syndrome. The Journal of Pediatrics. 2014;164(6):1475-1480. https://doi.org/10.1016/j.jpeds.2014.01.056
31. Bonnet D, Cormier-Daire V, Kachaner J, *et al.,*Microsatellite DNA markers detect 95% of chromosome 22q11 deletions. Am J Med Genet 1997; 68:182-4.
32. Peyvandi S, Lupo PJ, Garbarini J, *et al.,*22q11.2 deletions in patients with conotruncal defects: data from 1,610 consecutive cases. Pediatric Cardiology. 2013;34(7):1687-1694. https://doi.org/10.1007/s00246-013-0694-4
33. N. Philip, R. Reynaud. Hypocalcemia and 22q11.2 microdeletion. Archives de Pediatrie 2008;15: p648-p649. (c) 2008 Elsevier Masson SAS.
34. Ariane JM, Jamil A. Perloff's Clinical Recognition of Congenital Heart Disease, 7th Edition. Elsevier, 2022. ISBN: 9780323529648
35. Thankavel PP, Brown PS, Nugent AW. High-takeoff single coronary artery with intramural course in truncus arteriosus: prospective echocardiographic identification. Pediatr Cardiol. 2013; 34:1517-9.
36. Mittal K, Dey AK, Gadewar R, *et al.,* rare case of truncus arteriosus with anomalous origin of the right coronary artery from the pulmonary artery (ARCAPA) and unilateral left pulmonary artery agenesis. Jpn J Radiol. 2015; 33:220-4.
37. Alboliras ET, Lombardo S, Antillon J. Truncus arteriosus with double aortic arch: two-dimensional and color flow Doppler echocardiographic diagnosis. Am Heart J. 1995;129(2):415-7.
38. Jacobs ML. Congenital Heart Surgery Nomenclature and Database Project: Truncus Arteriosus. Ann Thorac Surg 2000;69: S50-55
39. Alejandro R. Clinical findings in common arterial trunk. Progress in Pediatric Cardiology 15 (2002) 23-31. Elsevier.
40. Chan FP. Computed Tomography and Magnetic Resonance Imaging in Neonates with Congenital Cardiovascular Disease. 2015 Elsevier. http://dx.doi.org/10.1053/j.sult.2015.01.006
41. Rice MJ, Seward JB, Hagler DJ, Mair DD, Tajik AJ. Definitive diagnosis of truncus arteriosus by two-dimensional echocardiography. Mayo Clin Proc 1982; 57:476-81.

42. Yoo SJ, Lee YH, Kim ES, *et al.,* Three vessel view of the fetal upper mediastinum: an easy means of detecting abnormalities of the ventricular outflow tract and great arteries during obstetric screening. Ultrasound Obstet Gynecol 1997; 9:173-82.
43. McElhinney DB, Reddy VM, Tworetzky W, *et al.,* Early and late results after repair of aortopulmonary septal defect and associated anomalies in infants less than 6 months of age. Am J Cardiol. 1998;81(2):195-201
44. D. Luu A. Aortopulmonary window. EMC Surgical Techniques - Thorax [42-765]. Doi:10.1016/S1241-8226(14)64610-7
45. M-L Hsieh. Single origin of right and left pulmonary arteries from ascending aorta with atretic main pulmonary artery from right ventricle and left pulmonary sling
46. C. Lapierre. Tetralogy of Fallot: Preoperative assessment with MR and CT imaging. 2016 Editions francaises de radiologie. Doi: 10.1016/j.diii.2016.01.009
47. Reddy VM, Rajasinghe HA, McElhinney DB, Hanley FL. Performance of right ventricle to pulmonary artery conduits after repair of truncus arteriosus: a comparison of Dacron-housed porcine valves and cryopreserved allografts. Semin Thorac Cardiovasc Surg. 1995; 7:133-138.
48. Danielson GK, Anderson BJ, Schleck CD, Ilstrup DM. Late results of pulmonary ventricle to pulmonary artery conduits. Semin Thorac Cardiovasc Surg. 1995; 7:162-167.