The Natural History of Truncus Arteriosus

CARLO MARCELLETTI, M.D., DWIGHT C. MCGOON, M.D.,
AND DOUGLAS D. MAIR, M.D.

SUMMARY The cases of 23 patients whose condition was diagnosed as truncus arteriosus, type I or II, and who were seen at the Mayo Clinic during the decade preceding 1967, that is, before corrective operation became feasible, were reviewed. Ten were infants (through one year of age), and all ten have died. Eight ranged in age from more than one year through seven years of age, and all are living, except one, who died 11 years after diagnosis. Five were older than seven years, and all had severe pulmonary vascular obstruction.

THE PROGNOSIS FOR PATIENTS with truncus arteriosus is generally considered to be poor. The median age at death, reported in autopsy series, varies from a few weeks to 6 months. Congestive heart failure secondary to a large pulmonary blood flow, with or without associated truncal valve regurgitation, is the major cause of death in early infancy. In clinical series, 15 to 30% of patients had survived beyond the first year of life, but progressive pulmonary vascular obstructive disease or clinically significant truncal valve regurgitation or both had developed in many. Late follow-up studies of the natural history of truncus arteriosus have not been previously reported.

Both palliative and corrective operations are now available for the management of patients with truncus arteriosus. Therefore, it is of importance to analyze the natural history of this condition in order to define how significantly it might be altered by operative management. Such is the purpose of this study.

Clinical Material

Between October 1957 and September 1967 and before the advent of a definitive surgical repair of truncus arteriosus, 23 patients (eight males and 15 females) were referred to the Mayo Clinic for evaluation of congenital heart disease (tables 1, 2, and 3). The diagnosis of truncus arteriosus, type I or II, was confirmed by cardiac catheterization and angiocardiography, exploratory thoracotomy, or autopsy. (We did not encounter type III truncus arteriosus and prefer to use “pulmonary atresia” to designate the condition of patients previously classified as “type IV truncus arteriosus.”) The age at which the patient was first referred for evaluation varied from 8 days to 16 years. Eight patients presented with a history of both congestive heart failure and cyanosis, seven with congestive heart failure alone, two with cyanosis only, and six with no symptoms. Eighteen patients had been treated previously with digitalis. Pertinent data from cardiac catheterization and angiocardiography are listed in tables 1, 2, and 3. Moderate to severe pulmonary vascular obstructive disease (pulmonary resistance [Rp] equal to or greater than 8 U m\(^2\)) was present in ten of the 13 patients in whom complete hemodynamic data were obtained. Two patients had unilateral absence of a pulmonary artery and four had angiographic evidence of mild to moderate truncal valve regurgitation. Follow-up information to August 1975 was obtained for all but one of the survivors, either by examination at the Mayo Clinic or by information from the referring physician, the parents, or the patient, or from all of them in answer to a special questionnaire.

Results

Ten of the 23 patients (43%) were referred for evaluation of this cardiac defect before the age of two years; in the remaining 13 the age ranged between two and 16 years (median 7.4 years). Of the ten infants, only two survived beyond their second year of life; they died subsequently, 8.5 and 12 years after their initial examination, of congestive heart failure and acute respiratory infection, respectively. Four of the older patients died one to 15 years after their initial examination at this clinic. Thus, a total of 14 patients died: eight from acute intractable congestive heart failure at a median age of 19.9 months; one from intractable ventricular tachyarrhythmia after cardiac catheterization (8-month-old infant in congestive heart failure); one from acute respiratory infection who was essentially asymptomatic from the congenital cardiac defect; two of the 3 patients in whom pulmonary resistance was more than 20 U m\(^2\) died suddenly at 15 (case 22) and 24 years (case 19) of age.
respectively, and two patients died after prolonged progressive fatigue and cyanosis, suggesting progressive pulmonary vascular obstructive disease (tables 1, 2, and 3). All patients except two (cases 4 and 16) had type I truncus arteriosus. In two patients (cases 9 and 18) only the right pulmonary artery was present. Six specimens of the heart were available for re-examination (table 4). In three cases the semilunar cusps of the truncal valve were found to be grossly deformed—thickened and nodular—strongly suggesting inadequate coaptation in vivo and consequent truncal valve regurgitation. In one case (case 20) the postmortem histologic examination of the lungs performed elsewhere revealed diffuse thrombotic occlusion of the pulmonary arterioles.

No response to our present follow-up was obtained from one patient. Eight of the 22 patients with completed follow-up are living at the time of this writing, 7 to 15 years after the first Mayo Clinic examination, their ages ranging from 11 to 26 years (median 18.1 years). The late status of the 11 patients over one year of age in whom the level of pulmonary resistance was known at initial examination is summarized in table 5. The present hematocrit was determined in four and was elevated in each, although in two other patients the hemoglobin concentration was normal. Only one patient is presently asymptomatic.

Discussion

Correct decisions regarding the management of patients with congenital heart defects require knowledge of the natural history of the particular condition. A lack of documentation of natural history before the availability of surgical interventions has proved a significant handicap in the identification of accurate indications and timing for operation of most congenital cardiac defects.

The clinical course of patients with truncus arteriosus showed great variability.\textsuperscript{16, 17} Though some patients survived to middle age,\textsuperscript{16, 18} most died early in life.\textsuperscript{1} The present study of 23 patients having the diagnosis of truncus arteriosus established before the availability of a corrective operation, though a small sample, provides some interesting observations. With remarkable internal consistency, the patients are divisible into three groups, according to age at diagnosis:

\textbf{Group 1.} One year or less in age (cases 1 through 10). All ten patients have died; all but three died soon after diagnosis, and those three lived 1, 8.5, and 12 years, respectively. All presented with congestive heart failure except the one who lived 12 years, who was asymptomatic at the time of diagnosis. Pulmonary resistance had been calculated in only two patients, and in these it averaged 4.2 U m\textsuperscript{2}.

\begin{table}[h]
\centering
\caption{Data on Group 1 (10 cases)}
\begin{tabular}{|c|c|c|c|c|c|c|c|}
\hline
\textbf{Case} & \textbf{Sex, age at Dx} & \textbf{Symptoms} & \textbf{Systemic O\textsubscript{2} sat.}, & \textbf{Systemic arterial pressure, mm Hg} & \textbf{Rp. U m\textsuperscript{2}} & \textbf{Associated defects} & \textbf{Death} \\
\hline
1 & F, 8 da & CHF, C & 89 & — & — & — & 10 days CHF\textsuperscript{†} \\
2 & F, 2 mo & CHF & 90 & 85 & — & — & Few days CHF\textsuperscript{†} \\
3 & F, 2 mo & CHF & — & — & — & — & Few days CHF\textsuperscript{†} \\
4 & F, 3 mo & CHF & — & — & — & — & Few days CHF\textsuperscript{†} \\
5 & F, 5 mo & CHF & 80 & 80 & — & Mild TVR & 1\frac{1}{2} yr CHF \\
6 & F, 6 mo & CHF & 82 & 84 & — & PDA, ASD & 2 mo Ventricular\textsuperscript{†} &
\textbf{tachyarrhythmias}
\hline
7 & F, 6 mo & CHF & 92 & 73 & 1.4 & PDA, tricuspid regurg, LPA absent & 1 mo CHF \\
8 & M, 8 mo & CHF & 90 & — & — & Few days CHF\textsuperscript{†} \\
9 & F, 1 yr & CHF, C & 86 & 80 & — & PDA, atrial septal defect & 12 yr ARI \\
\hline
10 & M, 1 yr & Asympt & 91 & 88 & 7.0 & — & 12 yr ARI \\
\hline
\end{tabular}
\textsuperscript{*Where no value is given, the pulmonary artery pressure was unknown.} \\
\textsuperscript{†Autopsy performed at Mayo Clinic; specimen available.} \\
\textsuperscript{Abbreviations: CHF = congestive heart failure; C = cyanosis; TVR = truncal valve regurgitation; PDA = patent ductus arteriosus; ASD = atrial septal defect; ARI = acute respiratory infection; LPA = left pulmonary artery.}
\end{table}

\begin{table}[h]
\centering
\caption{Data on Group 2 (8 Cases)}
\begin{tabular}{|c|c|c|c|c|c|c|c|}
\hline
\textbf{Case} & \textbf{Sex, age at Dx} & \textbf{Symptoms} & \textbf{Systemic O\textsubscript{2} sat.}, & \textbf{Systemic arterial pressure, mm Hg} & \textbf{Rp. U m\textsuperscript{2}} & \textbf{Associated defects} & \textbf{Follow-up (after 1st exam)} \\
\hline
11 & M, 2 yr & C & 87 & 105 & 16.0 & — & Alive 14 yr Class II, Hct 55\% \\
12 & F, 2 yr & CHF, C & 84 & 85 & 11.6 & — & Alive 9 yr Class II, Hct 60\% \\
13 & F, 4 yr & Asympt & 89 & 82 & 8.0 & Mild TVR & Alive 15 yr Class III, Hct 64\% \\
14 & F, 4 yr & CHF, C & 77 & 104 & 11.3 & Mod TVR & Alive 7 yr Class II, Hct 50\%, digita lis \\
15 & M, 5 yr & Asympt & 89 & 80 & 5.4 & — & Alive 14 yr Class I, Hb 14.5 g/dl \\
16 & M, 7 yr & Asympt & 88 & 108 & — & Dead 11 yr (cause, CHF) \\
17 & F, 7 yr & Asympt & 97 & 112 & — & Alive 15 yr Class II \\
18 & F, 7 yr & CHF, C & 90 & 111 & 11.0 & ASD; LPA absent & Alive 1 yr Class II, Hb 14.6 g/dl (last FU in 1961) \\
\hline
\end{tabular}
\textsuperscript{*Where no value is given, the pulmonary artery pressure was unknown.} \\
\textsuperscript{Abbreviations: CHF = congestive heart failure; C = cyanosis; TVR = truncal valve regurgitation; ASD = atrial septal defect; LPA = left pulmonary artery.}
\end{table}
Table 3. Data on Group 3 (5 Cases)

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex, age at Dx</th>
<th>Symptoms</th>
<th>Systemic O₂ sat., %</th>
<th>Systemic arterial pressure, mm Hg</th>
<th>Rp, U m²</th>
<th>Associated defects</th>
<th>Follow-up (after 1st exam)</th>
</tr>
</thead>
<tbody>
<tr>
<td>19</td>
<td>M, 10 yr</td>
<td>CHF, C</td>
<td>67</td>
<td>102</td>
<td>27</td>
<td>PFO</td>
<td>Dead 14 yr; sudden death*</td>
</tr>
<tr>
<td>20</td>
<td>M, 10 yr</td>
<td>CHF, C</td>
<td>75</td>
<td>110</td>
<td>23</td>
<td>PDA, interrupted arch</td>
<td>Dead 1 yr; CHF*</td>
</tr>
<tr>
<td>21</td>
<td>F, 10 yr</td>
<td>Asymp</td>
<td>88</td>
<td>101</td>
<td>10</td>
<td>—</td>
<td>Alive 11 yr Class II</td>
</tr>
<tr>
<td>22</td>
<td>M, 14 yr</td>
<td>CHF, C</td>
<td>76</td>
<td>121</td>
<td>30</td>
<td>Mod TVR</td>
<td>Dead 1 yr; sudden death</td>
</tr>
<tr>
<td>23</td>
<td>F, 16 yr</td>
<td>C</td>
<td>87</td>
<td>103</td>
<td>19</td>
<td>—</td>
<td>Alive 10 yr Class II</td>
</tr>
</tbody>
</table>

*Autopsy performed elsewhere; specimen available.
**Abbreviations: CHF = congestive heart failure; C = cyanosis; PFO = patent foramen ovale; PDA = patent ductus arteriosus; TVR = truncal valve regurgitation.

Group 2. Older than 1 year through 7 years of age (cases 11 through 18). One died 11 years after diagnosis at age 18; the other seven patients are living. Only two presented with congestive heart failure. In six the Rp calculated had averaged 10.6 U m². All but one of the surviving seven patients have some degree of disability, and four of six have elevation of hematocrit or hemoglobin.

Group 3. Older than seven years of age (cases 19 through 23). Three of the five patients have died, and the remaining two are mildly symptomatic. The three who died presented with congestive heart failure, and the other two did not. The Rp had been calculated in all and averaged 21.8 U m².

Thus, eight patients died in infancy. The remaining six deaths occurred at ages 9 to 24 (average age, 15 years). Nine patients are still living, and in six of eight of these for whom data are available, the Rp was elevated on initial examination to a level considered to contraindicate operation.

The implications of these findings seem clear. The infant in cardiac failure due to truncus arteriosus is in urgent need of operative intervention. However, the operative manage-ment of the young infant in congestive heart failure is problematic. Banding of the pulmonary artery (or arteries) may provide effective palliation if the failure is secondary only to a large pulmonary blood flow but would not ameliorate the effects of any associated truncal regurgitation. There is evidence that significant truncal regurgitation is common in such infants.** In this study, deformities of the semilunar cusps suggesting truncal regurgitation were found in three of the six infants in whom autopsy was performed at this clinic. Furthermore, the clinical diagnosis of truncal regurgitation may not be precise, and even truncal root angiography may be misleading, since a large runoff into the pulmonary arteries may sometimes mask significant valvular regur-gitation.

Bandage itself is not without significant operative risk. In the series of Singh and colleagues,14 ten of 15 infants in intractable heart failure who underwent pulmonary artery banding for truncus arteriosus in the first year of life died of postoperative low cardiac output. One late death occurred, with an overall mortality of 73%, and autopsy showed truncal valve deformities in seven of 11 infants. As recently reviewed by Poirier and colleagues,19 this palliation carries

Table 4. Autopsy Findings

<table>
<thead>
<tr>
<th>Case</th>
<th>Age at death</th>
<th>Truncus arteriosus</th>
<th>Pulmonary blood supply</th>
<th>Truncal valve</th>
<th>Ventricular septal defect</th>
<th>Associated cardiac anomalies</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>18 days</td>
<td>Type II overriding entire RV</td>
<td>LPA from L lateral aspect of truncus</td>
<td>4 leaflets; cusps thickened &amp; nodular</td>
<td>In membranous portion of septum, posterosuperior to crista supraventricularis; reaching back to tricuspid annulus</td>
<td>Interrupted aortic arch, PDA, persistent L superior vena cava</td>
</tr>
<tr>
<td>2</td>
<td>2 mo</td>
<td>Type I equally overriding RV &amp; LV</td>
<td>Pulm artery from L lateral aspect of truncus</td>
<td>3 leaflets</td>
<td>Immediately beneath truncal valve; not reaching back to tricuspid valve annulus</td>
<td>Atrial septal defect</td>
</tr>
<tr>
<td>3</td>
<td>2 mo</td>
<td>Type I equally overriding RV &amp; LV</td>
<td>Pulm artery from L lateral aspect of truncus</td>
<td>3 leaflets</td>
<td>Immediately beneath truncal valve; not reaching back to tricuspid valve annulus</td>
<td>—</td>
</tr>
<tr>
<td>4</td>
<td>3 mo</td>
<td>Type II overriding RV &amp; LV slightly more right-sided</td>
<td>LPA &amp; RPA from posterosuperior aspect of truncus</td>
<td>2 leaflets</td>
<td>Immediately beneath truncal valve; not reaching back to tricuspid valve annulus</td>
<td>Atrial septal defect</td>
</tr>
<tr>
<td>7</td>
<td>8 mo</td>
<td>Type I equally overriding RV &amp; LV</td>
<td>Pulm artery from L lateral aspect of truncus</td>
<td>3 leaflets; free edges thickened &amp; curled</td>
<td>Immediately beneath truncal valve; not reaching back to tricuspid valve annulus</td>
<td>—</td>
</tr>
<tr>
<td>8</td>
<td>8 mo</td>
<td>Type I equally overriding RV &amp; LV</td>
<td>Pulm artery from L lateral aspect of truncus</td>
<td>4 leaflets; cusps thickened &amp; bearing warty red nodules</td>
<td>Immediately beneath truncal valve; not reaching back to tricuspid valve annulus</td>
<td>—</td>
</tr>
</tbody>
</table>
mortality rates ranging from 33 to 100% and averaging approximately 67%.

Complete repair of truncus arteriosus in infancy during our early experience resulted in an excessive early mortality (83%). In patients over two years of age, a gratifying progressive reduction in the surgical risk has been achieved; in this age group, the current operative mortality is about 9%. It is appropriate, therefore, that renewed attempts at corrective operation for the infant in cardiac failure due to truncus arteriosus are now indicated, with guarded optimism that a substantial number will thereby live.

Specific criteria exist for the selection of patients with truncus arteriosus for corrective operation, and elevated pulmonary resistance is the primary contraindication. In truncus arteriosus there appears to be an accelerated tendency toward pulmonary vascular obstructive disease. Because of this, corrective operation should be considered at about two years of age for the child with truncus arteriosus who has survived infancy. Such a policy may result in the necessity to replace most of the conduits between the right ventricle and pulmonary artery with a larger conduit some ten years or so later. Such replacement of conduits seems well tolerated since all of our 22 procedures for replacement of an aortic homograft conduit have been successful.

On the basis of our present knowledge of the early and late risk of surgical repair of truncus arteriosus, it is not conclusive whether the natural history of this condition has been significantly altered by operative management. In fact, the patients shown to be ideal candidates for repair are those between 5 and 12 years of age with Rp of < 10 U m². It is these very patients who appear to have a prolonged natural life expectancy. However, the significant reduction in operative mortality to 9% since 1972 for the corrective operation, the encouraging late status of patients surviving operation, and the grave outlook for patients with truncus arteriosus, as substantiated in this study, all serve to justify the continued enthusiastic application of corrective operation by groups experienced in the management of complex congenital cardiac defects.

References

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Circulation. 1976;54:108-111
doi: 10.1161/01.CIR.54.1.108
Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

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