Introduction

There are some discrepancies about accompanying anomalies. In several clinical and autopsy studies considering HLHS, extracardiac anomalies and/or genetic disorders that could affect the survival of infants were noted in up to 37% of the infants.

Methods

Retrospective study of fetuses with HLHS referred as aortic atresia with mitral atresia or hypoplasia and critical aortic stenosis evolved to a severely hypoplastic left ventricle diagnosed prenatally between 2012 and 2017 in a referral center.

Results

HLHS was found in 9.7% of fetuses with cardiovascular abnormalities (CVA). As an isolated anomaly was present in 40% of cases; in 24.5%, other CVA were detected; in 14%, CVA and extracardiac anomalies and in 21.5% only extracardiac malformations were present. Genetic disorders were detected in 18.4% (Turner syndrome in 5 cases, trisomy 13 in 2 cases, trisomy 18 in 2 cases, microdeletion 22q11.2 in 2 cases and in one case 9p23 deletion).

In three newborns, dysmorphic features were postnatally found but the female karyotypes were normal. The most common CVA was ventricular septal defect (40%). The most common noncardiac abnormalities were craniofacial and CNS defects and hydrops occurring at a frequency of 26%.

Conclusion

A diagnosis of HLHS is an indication for a detailed examination of cardiac and noncardiac structures. It is advisable to consider genetic testing, together with the microarray assessment. We showed a higher incidence of anomalies associated with HLHS than previously reported, and some of them are detectable only after birth.

Three cases of facial dysmorphia as well as squinting, blindness, deafness, tubular disorder and cryptorchidism were detected postnatally, accounting for 25% of extracardiac anomalies. Furthermore, 50% of cardiovascular anomalies were diagnosed postnatally (8 cases of VSD, 2 cases of ASD and 1 case of partial anomalous pulmonary venous return).