STILLBIRTH DUE TO UNRECOGNIZED CARDIAC ANOMALY: A FORENSIC AUTOPSY CASE REPORT

Ganesh Raj S[1], Norliza I[1], Mansharan Kaur CS[2,3].
[1] Department of Forensic Medicine, Sarawak General Hospital, Jalan Hospital, 93586 Kuching, Sarawak
[2] Institute of Pathology, Laboratory and Forensic Medicine (I-PPerForM), Universiti Teknologi MARA (UiTM), Jalan Hospital, 47000, Sg Buloh, Selangor, Malaysia
[3] Faculty of Medicine, Universiti Teknologi MARA, Jalan Hospital, 47000 Sg Buloh, Selangor, Malaysia

Correspondence:
Associate Professor Dr. Mansharan Kaur d/o Chainchel Singh
Institute of Pathology,
Laboratory and Forensic Medicine (I-PPerForM),
Level 4 Academic Building Universiti Teknologi MARA,
Sungai Buloh, Jalan Hospital,
47000 Selangor, Malaysia
Phone number
Email: mansharan@uitm.edu.my, mansharan70@gmail.com

Abstract

Congenital heart disease (CHD) is the result of a complicated interplay between genetic and non-genetic, or "environmental," factors acting on the foetus and one of those environmental factors is maternal hyperglycemia. Maternal diabetes has teratogenic effects on the evolution of the foetal cardiovascular system; as a consequence, cardiovascular malformations are the most common anomalies in infants of diabetic mothers with transposition of the great arteries, tricuspid atresia and truncus arteriosus being some of the common cardiac malformations encountered. Thus, it is important to perform a detailed heart examination at autopsy of perinatal deaths in order to ascertain related anomalies. We present a case of stillbirth in a woman with Type 1 Diabetes mellitus on insulin therapy who claims she was unaware about her pregnancy.

Keywords: *Congenital Heart Disease, Maternal Diabetes, Teratogenic, Transposition of the Great Arteries*

Introduction

Maternal insulin–dependent diabetes is highly associated with congenital malformation affecting embryo development, with fatal malformations occurring in 2.1% of infants born to diabetic mothers as compared to 0.3% in infants of non-diabetic mothers. These congenital malformations, however, do not appear to be solely genetic in origin since the number of malformed offspring is not increased in diabetic fathers (1).

Though cardiac anomalies commonly occur in foetuses that are exposed to environmental agents such as rubella virus, drugs, alcohol and genetic causes such as Trisomy 18 and Goldenhar Syndrome, there is also a strong association between maternal diabetes and the absolute risk for central nervous system, cardiovascular and other major birth defects with transposition of the great arteries being one of the commonest congenital heart anomalies, accounting for 3% of all congenital heart defects (2, 3).

Transposition of the great arteries is a condition where the aorta arises from the right ventricle rather than the left ventricle and the pulmonary artery arises from the left ventricle instead of the right ventricle, leading to de-oxygenated blood being carried back to the body and oxygenated blood pumped back to the lungs instead of the body.

External marks of injuries due to beating, shaking, throwing or dropping of the foetus must also be ruled out beforehand since these injuries are useful indicators in differentiating non-accidental injuries (NAI) from accidental injuries, as literature suggests a higher risk of NAI in children born prematurely or with multiple medical conditions (4).

Thus, these potential biases are vital in determining the cause of death and facilitating investigation.

Case Presentation

We present a case of a 23-year-old married woman with Type 1 Diabetes mellitus on insulin therapy who experienced severe lower abdominal cramps on the 20th of January 2019 around 1215 h. She was alone at home and thus was unable to seek medical attention immediately. She

eventually passed out a foetus at home and sought medical attention the following morning claiming she was unaware of this pregnancy as her menstrual cycles were regular with the last cycle being in December 2018, lasting for 5 days.

A postmortem examination done revealed a male foetus with a body weight of 715 gm, crown heel length of 32 cm, crown rump length of 22 cm, head circumference of 22 cm, abdominal circumference of 19 cm and chest circumference of 21 cm. The estimated gestational age of the foetus was about 6 months according to *Haase Rule and quantitative standard for fetal and neonatal autopsy* (5). There was no evidence of dysmorphism, low set ears or spina bifida. There were also no abnormalities or injuries seen externally.

Both conjunctivae were congested but no petechial haemorrhage was seen. There was no skull vault fracture, subgaleal, subdural or subarachnoid haemorrhage. Both lungs were collapsed and the heart weighed 5 gm. Further examination of the cardiovascular system revealed transposition of the aorta and pulmonary artery. The aorta was seen to arise from the right ventricle (Figure 1), and the pulmonary artery from the left ventricle (Figure 2). There was no atrial or ventricular septal defect. All other organs were in normal anatomical position with no anomalies.

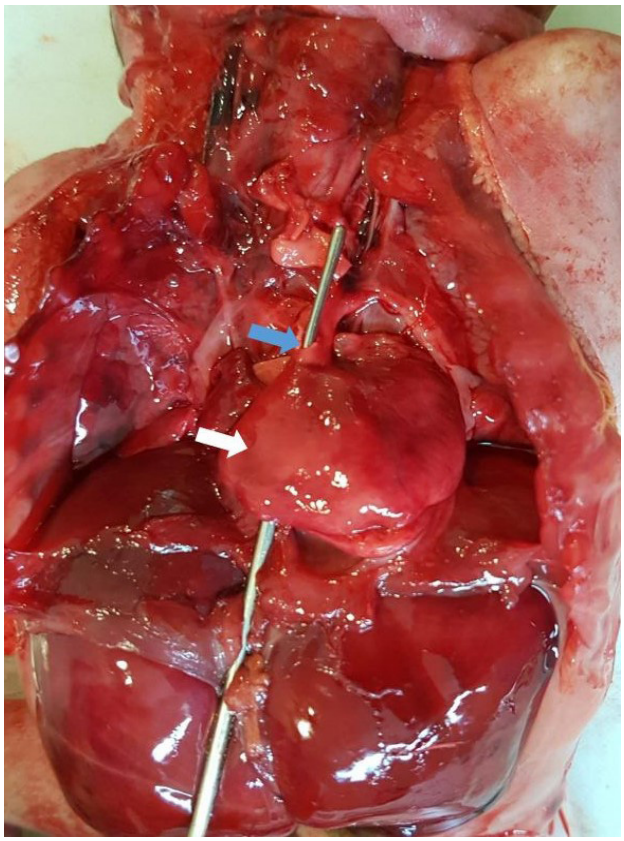

Figure 1: Transposition of the great vessels showing aorta (blue arrow) arising from the right ventricle (white arrow)

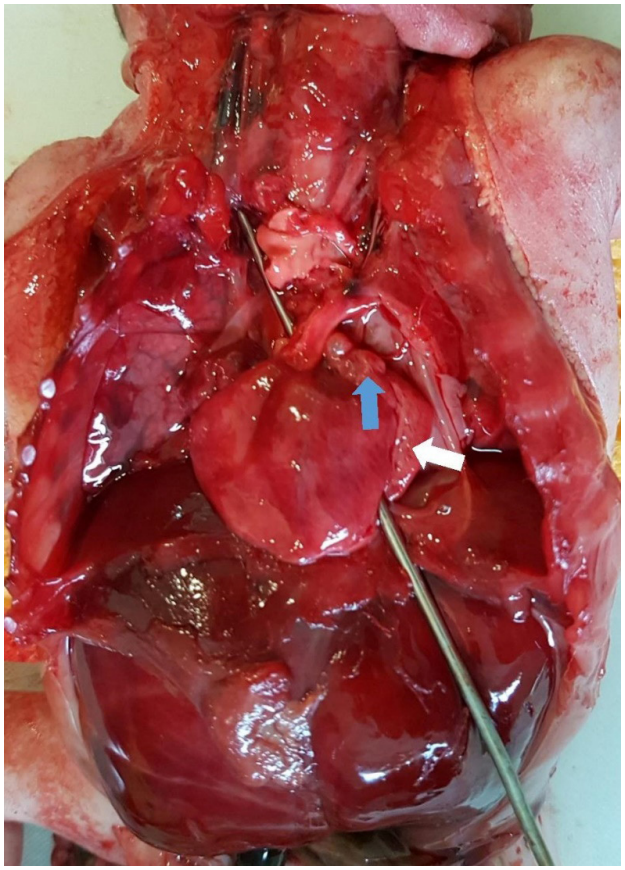

Figure 2: Transposition of the great vessels showing pulmonary artery (blue arrow) arising from the left ventricle (white arrow) aorta (blue arrow) arising from the right ventricle (white arrow)

The placenta weighed 505 gm and the umbilical cord measured 41 cm with 1 vein and 2 arteries. There was no evidence of petechial haemorrhage on the maternal or foetal surface of the placenta and the cotyledons were normal.

However, blood investigation results from the mother's medical records showed poor glycaemic control (Table 1). It was thus concluded that the cause of death was stillbirth due to transposition of great arteries in an infant of a diabetic mother.

Table 1: Maternal blood investigation results

Blood Parameters	January 2018	November 2018
HbA1C	9.4%	8.8%
Fasting Plasma Glucose	8.0 mmol/L	8.2 mmol/L
2-Hour Post-Glucose Load	11.8 mmol/L	11.4 mmol/L

Discussion

Maternal insulin-dependent diabetes has long been associated with congenital malformations and literature states that fatal malformations are approximately six times more frequent in infants of these mothers. Common congenital malformations documented involve the central nervous system, musculoskeletal, genitourinary and cardiovascular system with congenital heart anomalies being the most frequent malformation accounting for 21% of all anomalies (2, 5, 6).

The most common congenital heart anomalies encountered include ventricular septal defect, transposition of great arteries and aortic stenosis. Transposition of the great arteries (TGA) is one of the most common and severe paediatric cardiac congenital defects, arising from an embryological discordance between the aorta and pulmonary trunk (7). It is also called "blue-baby syndrome" due to the low amount of oxygen provided to the body and is a very specific congenital heart anomaly without additional extra cardiac anomalies as seen in this case (4). Heart anomalies such as atrial or ventricular septal defect provide some oxygen to the body because oxygen-poor and oxygen-rich blood mixes through the septal defects. However, without such heart anomalies mentioned, this condition will be fatal to the growing foetus, leading to miscarriages and stillbirths as evident in this case (5).

The pathophysiology of high stillbirth rates due to congenital malformations amongst pregnancies complicated by diabetes is unknown, but likely to be multifactorial with possible mechanisms including chronic hypoxia, acidosis and hyperglycaemia-induced teratogenesis. However, more studies are required to determine early foetal loss rate, metabolic status during organogenesis and to assess the effect of diabetic control on malformation rates (1, 6, 8).

It must be remembered that in addition to maternal diabetes there are other factors known to predispose to stillbirths, i.e., maternal age of 18-29 affecting male foetuses, poor socio- economic status, maternal infections and maternal obesity (9, 10). Most stillbirths however are preventable with adequate antenatal care which provides a natural contact with healthcare providers through which requisite interventions can be facilitated in order to prevent, identify or treat medical conditions, genetic factors or infections identified during pregnancy.

Conclusion

In conclusion, the findings of the forensic autopsy facilitated the investigation, established stillbirth and its cause. Thus, forensic pathologists are encouraged to perform a detailed heart examination at autopsy of perinatal deaths in order to ascertain related anomalies.

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Informed Consent

Verbal consent for publication was obtained from the parents of the deceased and registered with the National Medical Research Register Malaysia (Research ID 48650).

Competing Interests

The authors declare that they have no competing interests.

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