Forensic Science International 254 (2015) e7–e12

Contents lists available at ScienceDirect

Forensic Science International

journal homepage: www.elsevier.com/locate/forsciint

Case report

Generalized arterial calcification of infancy—Findings at post-mortem computed tomography and autopsy

Ferdia Bolster *, Zabiullah Ali, Pamela Southall, David Fowler

Office of the Chief Medical Examiner, State of Maryland, 900 West Baltimore Street, Baltimore, MD 21223, USA

ARTICLE INFO

Article history:
Received 8 May 2015
Received in revised form 16 June 2015
Accepted 17 June 2015
Available online 26 June 2015

Keywords:
Generalized arterial calcification of infancy
Idiopathic infantile arterial calcification
Post mortem computed tomography
Autopsy

ABSTRACT

Generalized arterial calcification in infancy is a rare genetic disorder characterized by abnormal calcification of large and medium sized arteries and marked myointimal proliferation resulting in arterial stenosis. The condition is often fatal secondary to complications of cardiac ischemia, hypertension and cardiac failure. In this report we describe the findings at post mortem computed tomography, histology and autopsy.

1. Introduction

Generalized arterial calcification of infancy (GACI), also known as idiopathic infantile arterial calcification (IIAC) is a rare autosomal recessive genetic disorder characterized by abnormal calcification of large and medium sized arteries and marked myointimal proliferation resulting in arterial stenosis. The prognosis is extremely poor and 85% of affected infants die within the first 6 months of life [1,2]. Death is typically due to cardiovascular complications and heart failure [3,4]. This report describes the post mortem computed tomography (PMCT), histology and autopsy findings in a 3 week old infant.

2. Case report

A female baby was born at 37 weeks gestation following an uncomplicated antenatal course. She briefly required supplemental oxygen immediately after birth, but was discharged home from hospital 2 days later. At 22 days of life she was noted to be irritable overnight and the next morning developed respiratory distress with wheezing and grunting. She was noted to feel cold and stopped breathing. Emergency medical services were called, confirmed cardiopulmonary arrest, intubated the infant and commenced cardiopulmonary resuscitation. The infant remained asystolic and was subsequently pronounced dead in the emergency room.

Family history was noteworthy for a number of miscarriages and neonatal death to a maternal brother, and a history of several miscarriages to both maternal and paternal aunts. The infant underwent post-mortem computed tomography (PMCT) followed by conventional autopsy.

Radiologic and autopsy findings

PMCT scan was performed on a General Electric (GE) Light Speed RT-16 multi-detector (GE Healthcare, Milwaukee, Wisconsin, USA). Whole-body was scanned with a slice thickness of 0.625 mm and the acquired images were reconstructed in a contiguous fashion using the GE Advanced Workstation (AW-2) (Version: aws-2.0–5.5).

PMCT revealed extensive abnormal vascular calcification and sites of abnormal soft tissue calcification.

On the head CT portion there was high attenuation consistent with calcification affecting the pinna (cartilage) of the ears bilaterally (Fig. 1). There was no intracranial calcification, but vascular calcification was noted affecting the bilateral common and external carotid arteries, the vertebral arteries and several branches of the external carotid arteries.

On the whole-body CT scan there was extensive arterial calcification noted throughout the chest, abdomen and pelvic as well as both upper and lower limbs.

* Corresponding author at: 900 W Baltimore St, Baltimore, MD 21223, USA. Tel.: +1 410 333 6295; fax: +1 410 333 3063.
E-mail addresses: ferdiabolster@hotmail.com (F. Bolster), aliz@ocmemd.org (Z. Ali), southallp@ocmemd.org (P. Southall), fowlerd@ocmemd.org (D. Fowler).

http://dx.doi.org/10.1016/j.forsciint.2015.06.017
0379-0738/

Fig. 1. Coronal CT demonstrates bilateral ear cartilage calcification (white arrows). There is also calcification of the aortic arch (AA) and pulmonary artery (PA).

Within the chest the heart appeared enlarged, and there was bibasal lung atelectasis (Fig. 2). There was a correctly positioned endotracheal tube in situ, and there was extensive vascular calcification of thoracic and abdominal aorta and of the pulmonary arteries bilaterally (Fig. 3). The left main stem, left anterior descending, left circumflex and right coronary arteries were all heavily calcified (Fig. 4). Calcification was also noted along the branch vessels of the arch of the aorta, the subclavian arteries and their branches, and of the main upper limb arteries bilaterally (Fig. 3).

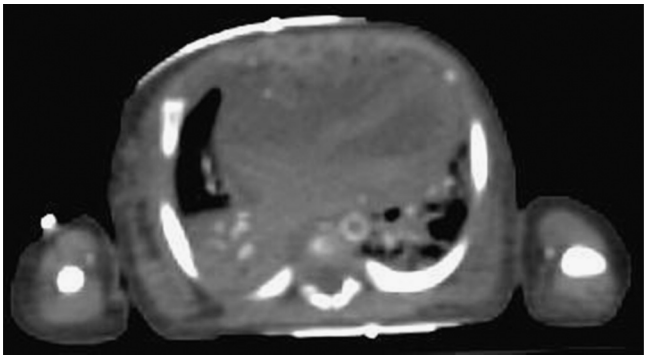

Fig. 2. Axial CT demonstrates cardiomegaly and bibasal atelectasis. There is also calcification of coronary arteries, aorta, pulmonary artery and upper limb arteries bilaterally.

Within the abdomen there was calcification in the walls of the abdominal aorta, celiac trunk, splenic, hepatic, renal, adrenal, superior mesenteric and common, lumbar, internal and external iliac arteries. There was further abnormal vascular calcification in the bilateral common and superficial femoral arteries, the proximal profunda femoris arteries and tibioperoneal trunks bilaterally.

Additionally there was also abnormal peri-articular soft tissue calcification at the left ankle joint (Fig. 5).

At autopsy, the external examination showed a well-developed and well-nourished neonate that weighed 2500 g, a crown-heel length of 51.8 cm, a crown-rump length of 36.8 cm, a head circumference of 35 cm and measured 45 cm in length. There was no evidence of trauma or dysmorphic features noted on the external examination.

Internally, the heart had a globoid configuration (Fig. 6A and B) and was enlarged at 42 g (normal 26 g, upper limit 40 g). All chambers were dilated. The left ventricular cavity diameter was 15 mm, left ventricular free wall thickness 5 mm, ventricular septum 6 mm, and right ventricular wall thickness 2 mm. The relationship between chambers and vascular connections were appropriate. Microscopically, there was biventricular circumferential subendocardial acute infarction with wavy hypereosinophilic myocardial fibers and interstitial acute inflammatory

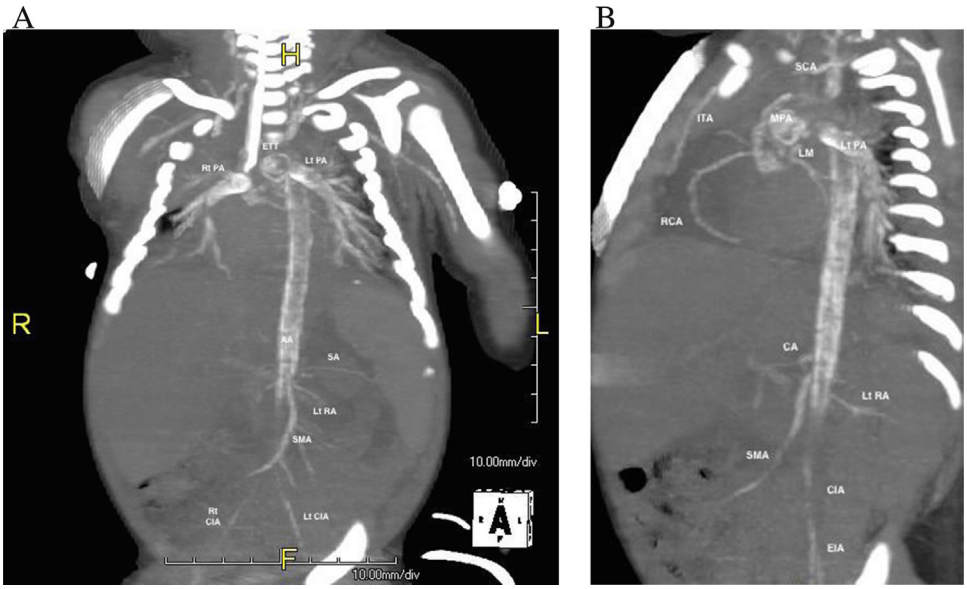

Fig. 3. (A and B): Coronal maximum intensity projection (MIP) CTs demonstrate an endotracheal tube (ETT) and extensive vascular calcifications (PA–pulmonary artery, SA—splenic artery, RA—renal artery, CIA—common iliac artery, BA—brachial artery, CA—celiac axis, SMA—superior mesenteric artery, SCA—subclavian artery).