Paediatric Critical Care Society

1398

ARTERIAL FUNCTION IN PREADOLESCENT CHILDREN FOLLOWING KAWASAKI DISEASE: A SYSTEMATIC REVIEW

¹Ioanna Zimianiti, ¹Paulina Cecula, ¹Carmen Traseira Pedraz, ²Malaz Elsaddig, ²Sundar Sathiyamurthy, ³Roshni Mansfield, ⁴Jayanta Banerjee. ¹Imperial College London School of Medicine; ²Department of Neonatology, Queen Charlotte's and Chelsea Hospital, Imperial College Healthcare Trust; ³Department of Neonatology, Queen Charlotte's and Chelsea Hospital, Imperial College Healthcare Trust; Biomedical Research Centre, Imperial College London; ⁴Department of Neonatology, Queen Charlotte's and Chelsea Hospital, Imperial College Healthcare Trust; Biomedical Research Centre, Imperial College London; Institute of Reproductive and Developmental Biology, Imperial College London; Origins of Child Health and Disease, Centre for Paediatrics and Child Health, Imperial College London

10.1136/archdischild-2021-rcpch.615

Background Kawasaki disease (KD) is a paediatric systemic vasculitis, which may alter arterial structure and function and predispose affected patients to long-term cardiovascular sequelae. Increased arterial stiffness is an independent predictor of cardiovascular morbidity and mortality and could be used as a biomarker of cardiovascular risk in KD patients.

Objectives To systematically review available literature in order to understand whether KD impacts arterial stiffness.

Methods This review was performed following PRISMA guidelines. PubMed was searched using the terms: 'pulse wave velocity' (PWV), 'carotid intima-media thickness' (cIMT), 'arterial stiffness index' (SIx), 'flow-mediated dilation' (FMD), 'flow imaging', 'laser flow Doppler', 'venous plethysmography', 'cardi* magnetic resonance imaging', 'aortic intima-media thickness', 'vascular ultrasound' and 'neonat*', 'paediatric', 'infant*', 'child*'. Case reports, case series, reviews, commentaries, conference proceedings, animal studies, articles not in English and articles with children >12 years old were excluded. Reference lists of relevant studies were searched for additional eligible studies. Studies assessing arterial structure and function in children affected by KD were included.

Results 12/1087 studies identified were included. Brachio-radial (brPWV) and aortic PWV (aPWV) were performed as measures of arterial stiffness in four studies, using techniques including echocardiography, photoplethysmography and magnetic resonance imaging. Five studies assessed cIMT and four studies measured flow-mediated dilation (FMD), as arterial wall structure and endothelial dysfunction can both affect arterial stiffness. Eight studies assessed biophysical properties of the aorta relating to stiffness (such as SIx and distensibility).

brPWV and aPWV were significantly higher in children with KD than healthy controls in all studies. This was independent of the presence of coronary artery lesions (CALs) in all but one study, which suggested a dose-response relationship between coronary involvement and brPWV. cIMT and SIx were also increased in children with KD except in one study. Children with KD had lower aortic distensibility than controls, and this was even lower in children with CALs compared to those without (0.6 ± 0.2 vs 0.9 ± 0.3 $\text{cm}^2/\text{dyne} \times 10^{-4}$, $p=0.001$).

All four studies showed FMD was impaired in children with KD, and most impaired in children with CALs or longer fever duration, suggesting a potential role of the endothelium in the pathogenesis of the disease. FMD improved with Pravastatin or intravenous Vitamin C treatment. CRP +1444 and TNF- α -308 polymorphisms were associated with higher carotid artery SIx and cIMT in patients with KD but not controls,

suggesting a genotype-modulating effect on the pro-inflammatory state that may mediate the increased risk of developing higher arterial stiffness in KD.

Conclusions Clearly, arterial stiffness measurements are increased in patients with KD, though the effect of presence of CALs on stiffness is yet to be conclusively determined. Variability between studies can be explained by the wide variety of techniques, patient demographics, and time from disease onset studied. An improved understanding of the pathophysiological mechanisms behind increased arterial stiffness may allow more targeted interventions to mitigate future cardiovascular risk. We recommend long-term cardiovascular monitoring of patients with KD as well as further longitudinal observational and interventional studies aimed at better understanding and offsetting the long-term arterial sequelae of KD.

British Society of Paediatric Endocrinology and Diabetes

1399

NEW ALPL GENETIC ALTERATION ASSOCIATED WITH AN ODONTOHYPOPHOSPHATASIA PHENOTYPE

¹Süleyman Yıldız, ²Funda Feryal Taş, ³Sabahattin Ertugrul. ¹Mardin Derik State Hospital; ²University of Health Sciences Diyarbakir Gazi Yaşargil Training and Research Hospital; ³Dicle University

10.1136/archdischild-2021-rcpch.616

Background Hypophosphatasia is a rare autosomal dominant/recessive metabolic disease characterized by defective bone mineralization as a result of defective alkaline phosphatase activity caused by mutations in the gene encoding the tissue nonspecific alkaline phosphatase (TNSALP) enzyme. Patients with isolated dental manifestations, typically presenting as premature loss of primary teeth, are classified as having odontohypophosphatasia. A subset of patients diagnosed with odontohypophosphatasia in childhood can later develop extra-dental manifestations that constitute childhood- or adult-onset hypophosphatasia.

Objectives We aim to share the clinical features of a patient diagnosed with odontohypophosphatasia which has a novel mutation and contribute to an increase in the awareness of this disease group.

Methods In the genetic analysis, a homozygous c.214A>Gp.172V mutation was found in the ALPL gene. According to the literature, this mutation had not been reported before. Heterozygous c.214A>Gp.172V mutation was found in the ALPL gene of the mother and father.

Results A 14-year-old girl was admitted to our hospital because of tooth deformity and discoloration. The patient's body weight was 51 kg (−0.5 SDS), and height was 154 cm (−1.04 SDS). According to the mother, the patient's milk teeth came out on time, but around the age of 3, her teeth began to fall off spontaneously. There was no similar complaint in the family of the patient. Somatic and motor development was normal and there was no walking problem. There was early loss of the anterior incisors due to insufficient mineralization in the teeth, lack or absence of cement on the tooth surface. The yellowish and brownish discoloration was also observed in the teeth.

In the biochemical examination, ALP: 14.2 U/L (40–150), Calcium: 10.1 mg/dl (8.4–10.2), Phosphor: 4 mg/dl (4–7), Magnesium: 1.93 mg/dl (1.3–2.1) PTH: 36.55 pg/ml (15–65),