and moderate pulmonary insufficiency.

The presentation showed sensorily child with multiple
eczematic lesions showed severely dry hands with
eczematous rash, paronychial drainage in the left
fingertip with a severe functional limitation (Figures 1, 2).

The pulmonary examination showed generalized tachypnea and tachycardia
with 152 bpm. A chest X-ray showed a pulmonary infiltrate of 94%, with room air saturation
of 96%. She was then admitted to the Pediatric Intensive Care Unit, where the
clinical parameters were monitored and periodic examinations were conducted.

The patient was referred to the Cardiology Department.

The patient was admitted to the intensive care unit with
a diagnosis of DC syndrome. The patient underwent a
surgical procedure to improve the heart function, which was
subsequently followed by DC syndrome.

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a diagnosis of DC syndrome. The patient underwent a
surgical procedure to improve the heart function, which was
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followed up by the palliative care unit. She died due to multiple organ failure nine months later.

Discussion

Multiple structural and functional alterations in the echocardiogram have been reported in RDEB patients including dilated aortic root, left ventricular hypertrophy, dilatation and dysfunction, increased left ventricular mass and depressed right ventricular systolic function.  

DC usually appears in cases of severe generalized RDEB. The most common EB subtypes associated with DC are RDEB and junctional EB-non-Herlitz.  

The age at diagnosis of DC in RDEB ranges from 2 to 28 years, with a variable ejection fraction from 9 to 45%. Mortality rate varies from 30-60% and death usually occurs within the first 3 months after diagnosis of DC. DC is usually detected at an advanced stage with low ejection fraction.  

Development of DC in RDEB is due to multifactorial disorders. Contributor factors may include chronic anaemia, iron overload, poor nutritional status, albumin and carnitine low levels, selenium and zinc deficiency, cardiotoxic drugs, viral infections, inflammatory conditions, structural defects in myocardium, chronic renal insufficiency and hypoaminoacidemia. To date there is no evidence of mutation in type VII collagen or the basement membrane zone in the pathogenesis of DC.  

History of prior viral infection is usually associated with a more favourable outcome. We suggest that our patient presented DC due mainly to chronic anaemia, iron overload and nutritional deficiency. Viral infection of the upper respiratory tract was the trigger for the development of heart failure and death.  

In conclusion, a RDEB patient who develops influenza-like illness and fever requires an urgent cardiac evaluation including electrocardiogram, chest radiograph and echocardiogram. Screening laboratory evaluations are also necessary including albumin, carnitine, selenium, zinc and haemoglobin levels and viruses serology. A complete drug history is essential. An early diagnosis and treatment can delay clinical progression and reduce morbidity and mortality.

Conflicto de intereses

Los autores declaran no tener ningún conflicto de intereses.

Bibliografía

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