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# Gorlin syndrome: Intracardiac tumor as initial manifestation

## Síndrome de Gorlin: tumor intracardiaco como manifestación inicial

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In the pediatric population, cardiac tumors are rare and may be associated with serious clinical conditions.

We present the case of a boy aged 3 years with a prenatal diagnosis of cardiac tumor detected by echocardiography (Fig. 1a) (Appendix A, Videos 1 and 2) and fetal MRI features compatible with cardiac fibroma. Prenatal genetic testing (array) for tuberous sclerosis turned out negative.

At 7 days of life, a cardiac MRI was performed, along with postnatal genetic testing (next-generation sequencing), which detected a mosaic de novo pathogenic variant in the *PTCH1* gene (c258_259del:p.Leu87IlefsTer2), leading to diagnosis of Gorlin syndrome.

Tumor growth and severe mitral insufficiency prompted performance of a partial resection at age 2 months. In the postoperative period, the patient developed pulsed ventricular tachycardia requiring electrical cardioversion and amiodarone. The condition was monitored with echocardiography (Fig. 1b–d) and Holter monitoring (Fig. 2) during the follow up, and the decision was made to implant a subcutaneous cardiac rhythm monitor at age 10 months.

The patient subsequently developed multiple basal cell carcinomas, confirmed by biopsy, and palmoplantar pits (Fig. 3). He is currently stable and free of arrhythmias with carvedilol and sacubitril-valsartan.

Gorlin syndrome is an inherited autosomal dominant disorder caused by variants of the *PTCH1* gene. It is characterized by the progressive development of numerous basal cell carcinomas, odontogenic keratocysts, skeletal abnormalities and multicentric tumors (intracranial, cardiac fibromas, ovarian).[1]

This syndrome should be considered in cases of cardiac tumor that are not consistent with tuberous sclerosis; early diagnosis through comprehensive genetic testing is key and monitoring of disease are key.[2,3]

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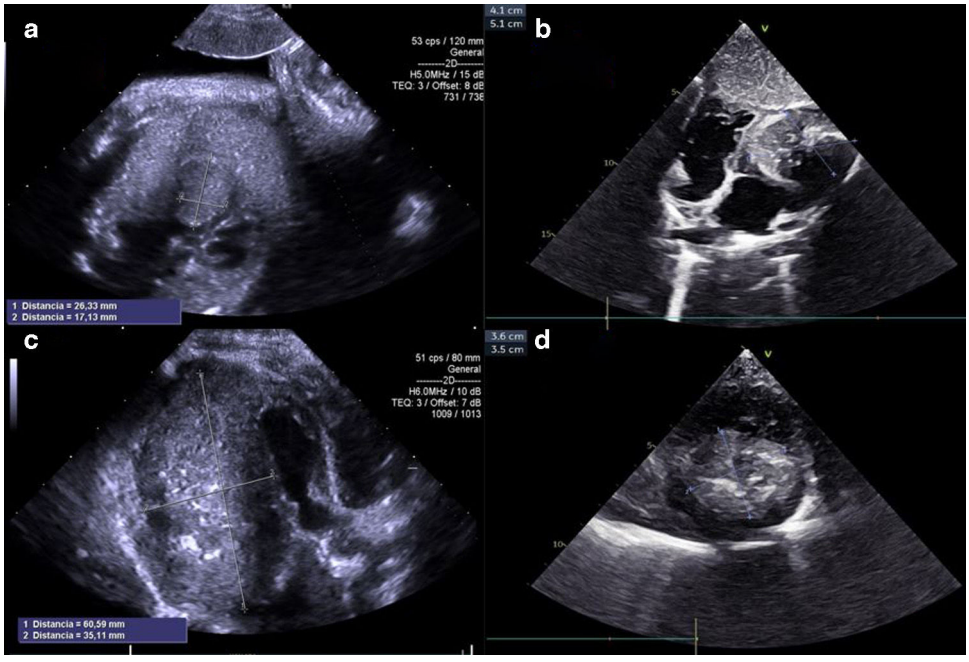


**Figure 1** (a) Fetal echocardiogram at $35^{+4}$ weeks of GA. Four-chamber view showing a $26 \times 17$ mm tumor in the left ventricle. (b) Echocardiogram following tumor resection at age 2 months, subcostal view ($41 \times 51$ mm). (c) Echocardiogram at age 4 months, parasternal long-axis view, intracardiac tumor ($60 \times 35$ mm). (d) Short-axis view showing tumor dimensions of $36 \times 35$ mm at age 3 years and 9 months.

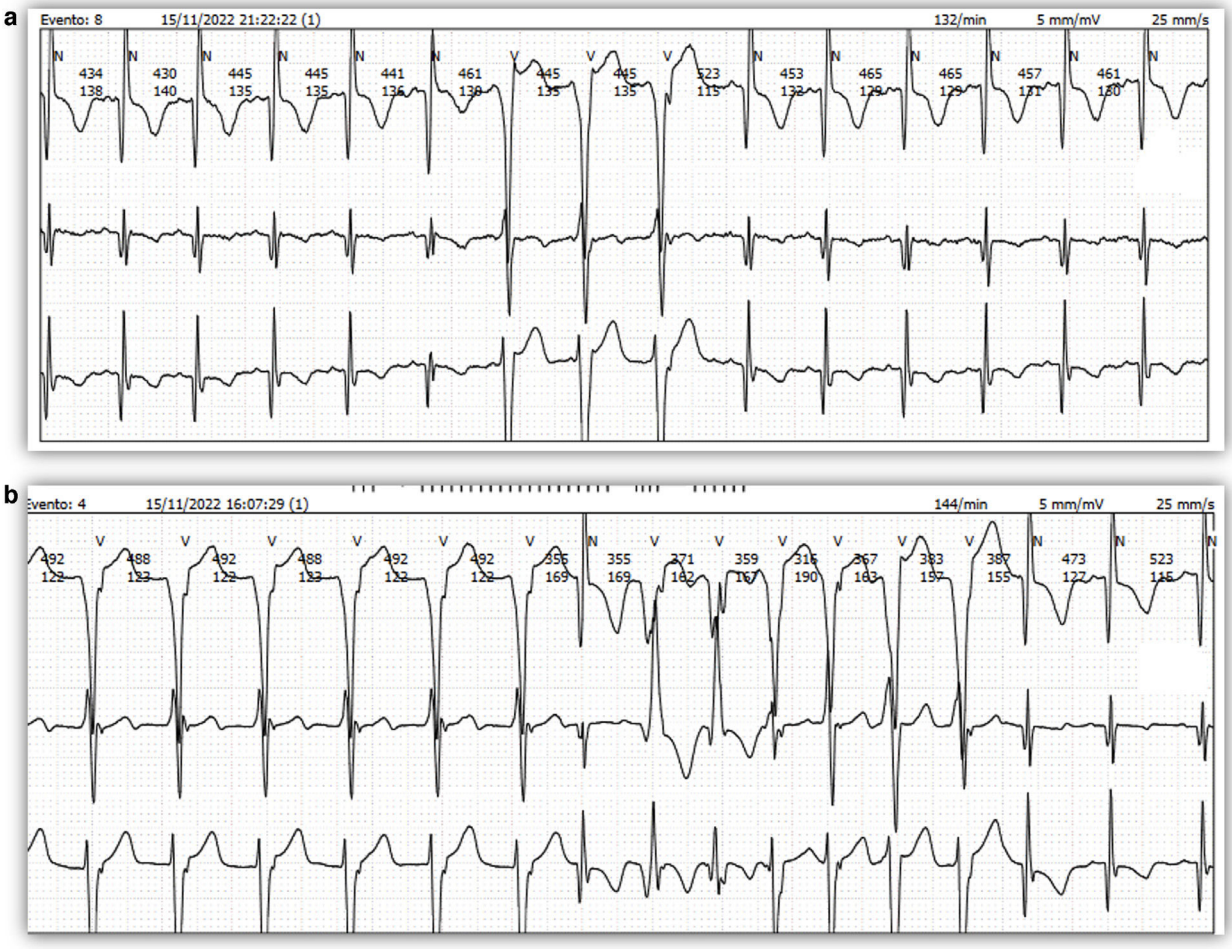


**Figure 2** (a) Holter tracing showing triplets. (b) Holter tracing showing run of nonsustained ventricular tachycardia.

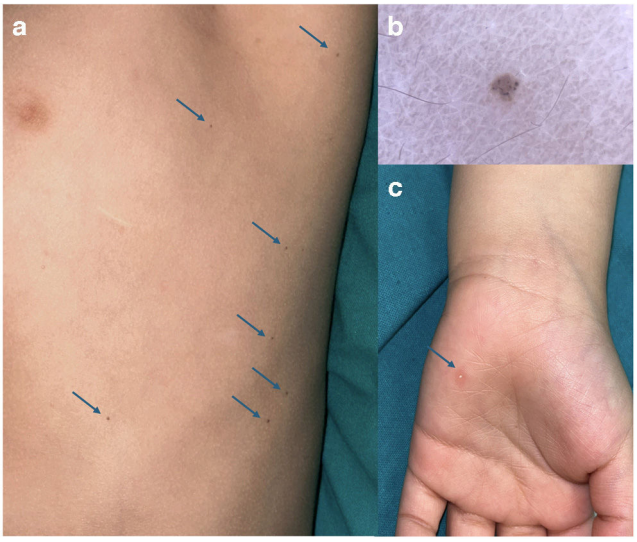


**Figure 3** (a) Multiple punctate brown papules located on the left side of the trunk, clinically and histologically consistent with basal cell carcinoma. (b) Dermoscopic image of one of the basal cell carcinomas showing blue-gray nests and globules. (c) Palmoplantar pit.

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## Declaration of competing interest

The authors have no conflicts of interest to declare.

## Appendix A. Supplementary data

Supplementary material related to this article can be found, in the online version, at doi:https://doi.org/10.1016/j.anpede.2026.504245.

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