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Brief Report

Clonal translocation in a cardiac fibroma presenting with incessant ventricular tachycardia in childhood

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THE AETIOLOGY OF SOFT TISSUE TUMOURS IS largely unknown, although chromosomal aberrations are found in some tumours,[1] suggesting that clonal translocation might play a causative role. Little is known of the cause of cardiac fibromas, but there has been one report of clonal translocation in this setting.[2] We report a case of a cardiac fibroma presenting with incessant ventricular tachycardia, where karyotyping of cells obtained at open biopsy of the tumour showed clonal translocation involving chromosomes 1, 9, and 5.

Case report

A 13-month-old girl presented in heart failure due to incessant ventricular tachycardia. She was treated with direct current cardioversion, amiodarone and captopril and her heart failure resolved although she continued to have intermittent, self-limiting episodes of tachycardia. Echocardiography showed poor ventricular function initially, which improved when sinus rhythm was restored, but there was a large mass in the apical part of the ventricular septum. Biochemical markers for embryonal tumours were negative and at open biopsy the tumour occupied the whole of the apex of the heart and was hard, with a smooth, white cut surface (Fig. 1).

Histology showed the tumour to be a fibroma with interweaving fascicles and whorls of spindle shaped cells in a stroma of collagen and elastin. Although the tumour was cellular, there were no features of anaplasia. Cytogenetic studies were carried out on the tumour tissue.

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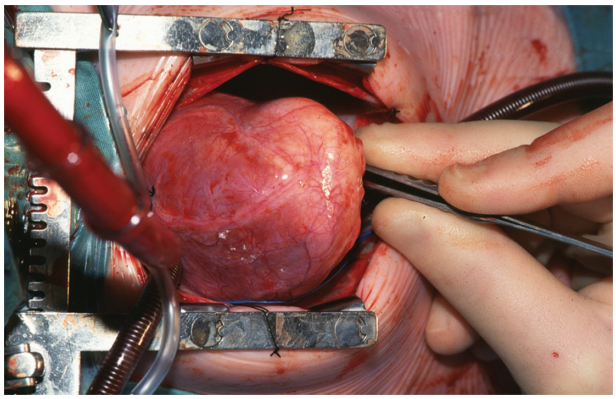

Figure 1.
Intra-operative photograph showing the location of the tumour at the apex of the ventricular mass.

Methods

Cell suspensions were obtained from fresh tumour tissue by mechanical and enzyme treatment using a modification of the collagenase digestion technique described by Limon et al.[3] Culture was established in a medium buffered with sodium bicarbonate and supplemented with 10% foetal calf serum, glutamine and antiobiotics. Chromosome preparations were made 12 days after culture initiation following exposure to colcemid (0.2 µg/ml) for 4 hours, followed by standard harvesting techniques. Analysis of the slides revealed an abnormal clone containing 46 chromosomes and a balanced t (1;9;5) translocation (Fig. 2). This is a 3-way reciprocal translocation between chromosomes 1, 9, and 5, with breakpoints at 1q42, 9q22 and 5q22 respectively. In short, 1q material has translocated to 9q, 9q material has translocated to 5q, and 5q material has translocated to 1q.

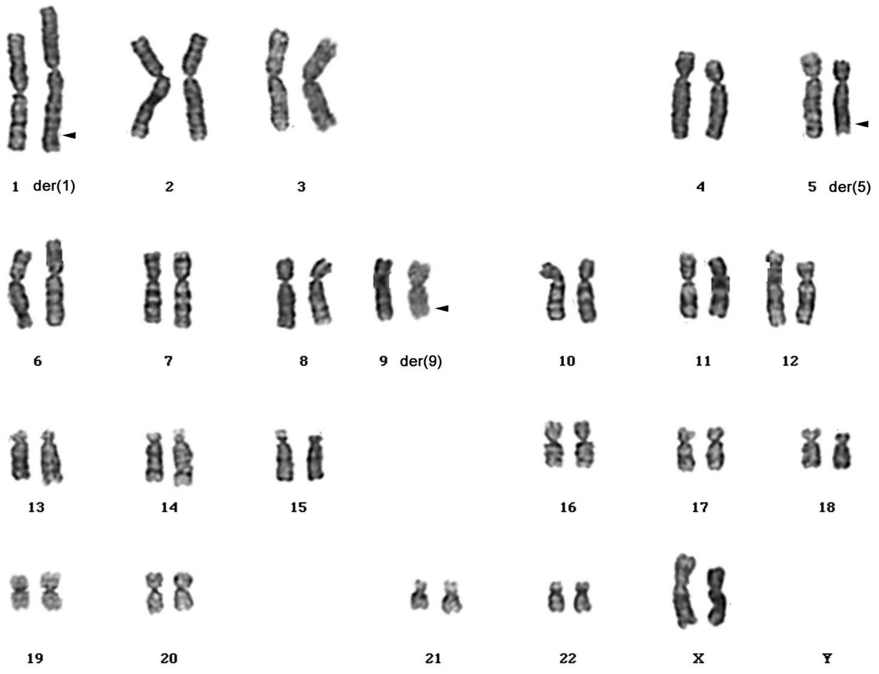

Figure 2.
Karyogram showing the translocation between chromosomes 1, 5 and 9 (arrowheads). "Der" is short for "derivative", and indicates the abnormal chromosome produced as a result of the translocation.

Discussion

Cardiac fibromas are connective tissue tumours, which constitute about one-tenth of cardiac tumours.[4] They are usually solitary, and are most often situated in the left ventricular myocardium or the ventricular septum. Cytogenetics has become an increasingly important tool in diagnosis and prognostication of such soft tissue tumours, and specific chromosomal aberrations have been reported in lipomas, leiomyomas, myxomas, schwannomas and sarcomas.[1] Little is known of the karyotype of fibroma cells, although one report suggests that specific abnormalities might also occur in these tumours.[2] The clonal translocation t (1;9;5) (q24;q22;q22) found in our patient appears to be a variant of that reported by Ferguson et al. suggesting that the t (1;9) translocation may be specific to cardiac fibromas. The exact mechanism of pathogenesis is yet to be elucidated, although it is likely that the creation of a chimeric fusion gene is involved. A vast amount of data about the cytogenetics of neoplastic disorders has accumulated in the past few decades, and several studies have shown that the presence of an abnormal clone may have prognostic significance.[5]

In view of these reports of specific translocations in cardiac tumours, and their potential diagnostic and prognostic implications, we recommend that karyotyping should become a routine part of the analysis of biopsies of cardiac tumours.

Acknowledgements

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