

Data supplement

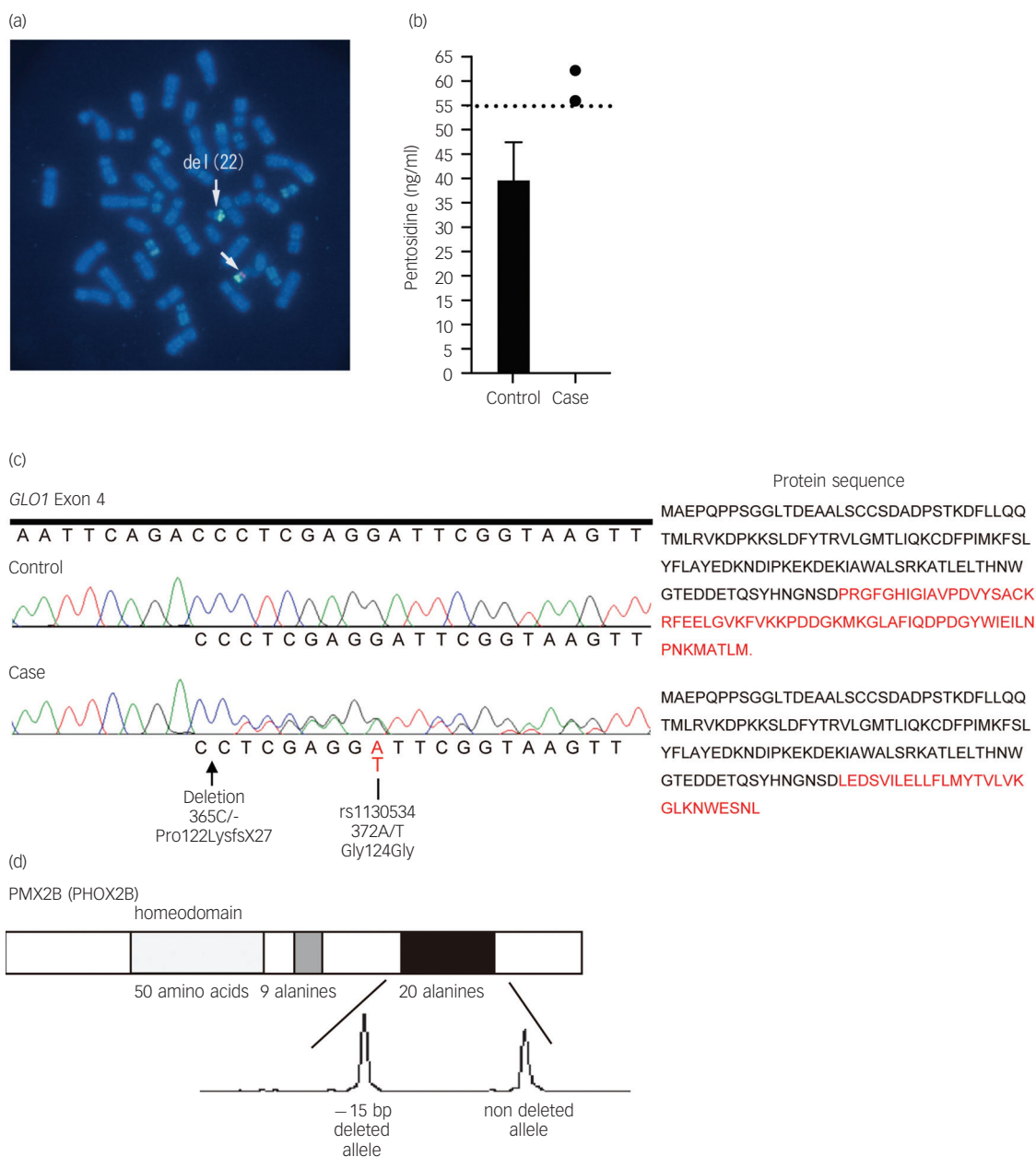


Fig. DS1 (a) Fluorescence *in situ* hybridisation; arrows mark both chromosomes 21 with the deleted 22q11.2 labelled. (b) Measurement of plasma pentosidine levels. The dotted horizontal line shows the mean + 2 standard deviation level of healthy controls. The pentosidine levels of our patient were measured twice, 1 week apart. (c) Sequencing results of *GLO1*. This shows a c.365delC in exon 4, resulting in a frameshift, and a silent polymorphism (rs1130534). The amino acid sequences of *GLO1* for both the control and the patient are shown to the right. (d) Structure of *PHOX2B* (upper panel) and results of fragment length analysis of a 20-alanine stretch portion in the patient, by ABI Gene Mapper.

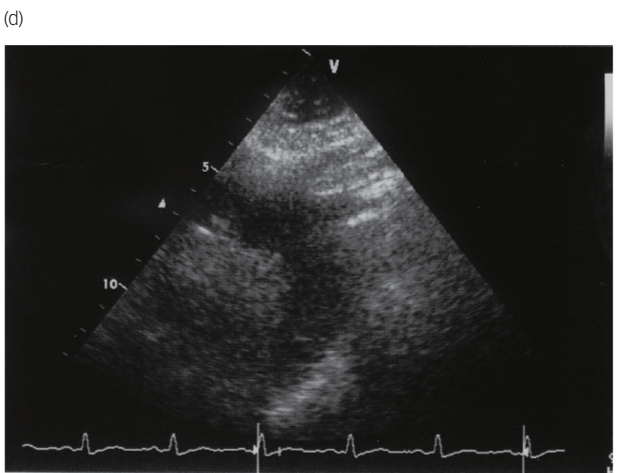
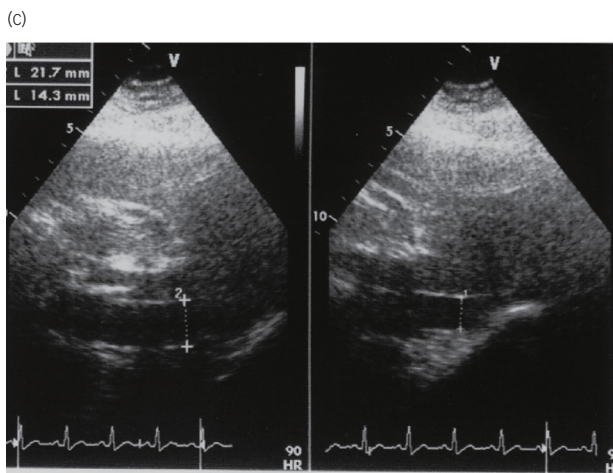
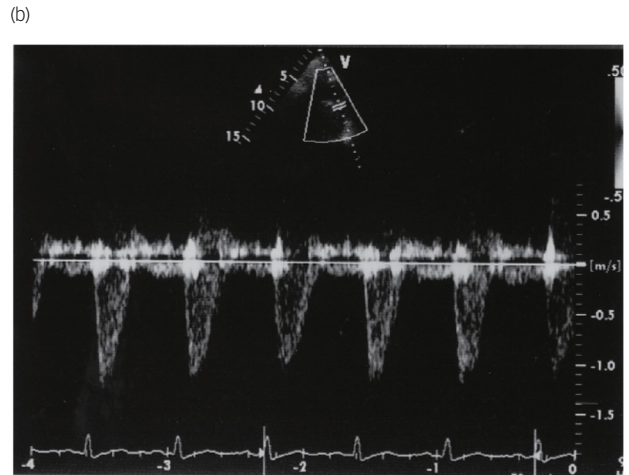
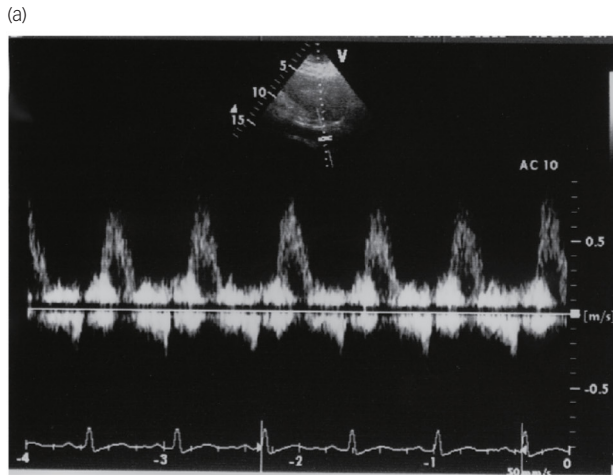


Fig. DS2 Echocardiograms: (a) abdominal aorta; (b) blood stream from aortic arc to descending aorta; (c) inferior vena cava; (d) aortic arc. There are no abnormalities in the cardiovascular system.