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.