The patient was a 38-year-old Caucasian man of Spanish origin.
His height was 165 cm and his parents were cousins.
Both parents were of normal height and neither showed any minor anomalies of the EvC syndrome spectrum.
The patient had two healthy sisters.
His morphological features were: (a) normal mouth opening with missing lower incisors (Fig.1); (b) small chest, as determined by a cardiothoracic examination; and (c) disproportionately short extremities with one additional postaxial digit on each hand (Fig.2).
Although no sensory-motor deficit was noted, the patient presented with mild mental retardation.
Genetic analysis in his early infancy showed a previously reported homozygous nonsense mutation c.1195 C1T, p.Arg399X of EvC2, resulting in loss of function of the protein.(5) His parents were heterozygous carriers of the mutation.
In 1998, the patient underwent successful ostium primum atrial septal defect closure through a midsternotomy.
He re-presented with embolic cerebral ictus in 2007, and cavotricuspid isthmus ablation for common atrial flutter was performed successfully.
There were no serious aftereffects.
During a routine examination in 2012, a high-pitched systolic murmur was discovered.
Electrocardiography showed sinus rhythm with hemiblock and right bundle branch block, while echocardiography revealed a dilated and hypertrophic left ventricle.
These abnormalities produced a flow that was directed to the left atrial appendage, leading to severe mitral valve regurgitation.
The patient's systolic pulmonary artery pressure was 50 mmHg and his aortic valve was normal.
His medical treatment included ramipril, duloxetine, alprazolam and acenocumarol.
The patient was scheduled for mitral valve surgery.
In the operating theatre, the patient's right femoral artery was cannulated.
A right thoracotomy was performed on the fourth intercostal space and bicaval cannulation was established.
Both veins were excluded.
The operation used a normothermic cardiopulmonary bypass without cross-clamping of the aorta in the beating heart.
To avoid air embolisms, continuous carbon dioxide (CO_2) field flooding was applied using a CarbonAid CO_2 diffuser (Cardia Innovation AB, Stockholm, Sweden).
The attempt to perform a mitral valve repair was unsuccessful and a bileaflet mechanical valve (no.29) was implanted.
No complications associated with anaesthesia occurred.
The patient's postoperative recovery was uneventful and he was discharged on postoperative day 10.
After two years of follow-up, echocardiography revealed optimal function of the mitral valve and a decrease in systolic pulmonary artery pressure (30 mmHg).