A three year three month old girl was admitted for assessment of developmental delay and dysmorphic facies. She was born in Greece, the only child of healthy, unrelated Greek parents at 32 weeks gestation, weighing 2.45 kg. The pregnancy was normal. Fetal movements were normal. At birth she was noted to have a broad forehead, hypertelorism, flat nasal bridge, large ears, long philtrum, micrognathia, loose skin and camptodactyly with thin, deep set nails. At three months she was noted to be a large child. The cry was low pitched and hoarse. She had a large ears, high arched palate and strabismus. There were prominent finger pads and narrow hyperconvex nails. There was mild kyphosis.