Proband was a 4-year-old girl from the Philippines. She was born at 39 weeks gestation, with a birthweight 1800g (−4SD). At four years of age, she weighed 4600g (−6SD), was 67cm in height (−10SD), with a head circumference of 37.2cm (−4.5SD). She had sensorineural hearing loss, a ventricular septal defect, and at age 4 was unable to walk or talk. She had arched eyebrows with long eyelashes, sparse scalp hair, a high palate, and posteriorly rotated ears with thickened helices. Her facial features seemed somewhat coarse and included upturned/anteverted nares, a long, smooth philtrum with thin upper lip, and wide downturned mouth.