Proband was a female infant born at 35 weeks gestational age weighing 3100g with a length of 50.8cm to non-consanguineous parents of Northern European descent. She was noted prenatally with ventricular septal defect, polyhydramnios and hydronephrosis, and postnatally had feeding difficulties requiring G-tube placement. Physical exam was significant for fine, arched eyebrows, long eyelashes, slight anteverted nares, broad nasal tip, a high, narrow palate with downturned corners of the mouth, a bifid uvula, somewhat coarse facial features, and retrognathia. She displayed bridged palmar creases bilaterally, as well as small 5th fingernails and absent 5th toenails. She died at 1 year of age after complications from a respiratory infection. A 610K SNP array was normal.