The proband was referred from a developmental assessment unit at the age of 11 because of hypnasal speech. Her parents are normal, white, and unrelated, and in their mid thirties. She has one sib, a female, two years older, who is normal. The pregnancy was uneventful and she was born at 37 weeks weighing 2340 g with a length of 48.8 cm. Her early development was slightly slow but not outside normal limits. She smiled at 12 weeks, sat at 8 months, crawled at 10 months, and walked at 15 months. She started to join words together by about 2 years of age. Ear infections started at 8 to 9 months. Glue ear was diagnosed and ventilating tubes were inserted at the first time at the age of 2 years. Feeding and swallowing have been unremarkable, though she occasionally regurgitated milk through the nose. An umbilical hernia was repaired at the age of 2 years. She is in mainstream schooling, but has needed remedial teaching. Teachers report an enthusiastic and cooperative girl who is having great difficulty with mathematics and any subject requiring logic reasoning. When tested on Stanford Binet IQ scale 4th edition, she functioned in the borderline to low average range.

On examination, at 11 years of age, her height was between the 10th and 25th centiles, weight between the 10th and 25th centiles, and head circumference between the 50th and 75th centiles. She had a “long” face, deficient alae, almond shaped palpebral fissures, dysplastic ears with intuned edges, and a small lower jaw (figure). There was no hypotonia. Serum calcium and parathryoid hormone were normal. Formal cardiac assessment and echo showed no abnormality, including the conotruncal region, such as right aortic arch. Immunological testing including T cell subsets, Concanavalin A, and T cell exterase were within normal limits. Her palate appeared short and she had hypnasal speech. Her speech was intelligible but the parents reported that often people ask her to repeat what she has said and her speech deteriorates when she is tired. Videofluoroscopy showed a good movement of the palate with a good knee to the palate; hearing was normal. Pharyngopalate and palatobounding resulted in reduction of hypnasality and increased intelligibility.

High resolution chromosome analysis to the 700 band level was normal. FISH analysis was performed using a cosmide probe corresponding to locus D22S75 (N25) and the DiGeorge critical region. A distal marker, Cos28, was used as a control probe to identify the chromosomes 22 homologues. A hybridisation signal was detected on only one of the chromosomes 22 in 15/15 metaphases examined with marker N25. This finding is consistent with the presence of 22q11 deletion.

Although this patient may represent one end of a spectrum, the absence of overt or submucous cleft palate, overt or subclinical conotruncal congenital heart disease, hypoparathyroidism, or thymic deficiency was not unusual in our 22q11 deleted patients. For the patient we describe here, the thymus may have failed to descend completely, but it is clearly functional. Further, it is clear that the size of the deletion does not appear to determine or correlate with the severity of the anomalies. Thus, ascertainment of mild cases in parents and their offspring is important for proper genetic counselling, as intramesial variability is common. This is an important syndrome for the dysmorphologist to recognise and, though con- 

Skeletmal malformations and polycystic kidney disease

Dr Winter’s comment on our paper describing an infant with autosomal dominant polycystic kidney disease (ADPKD) and skeletal malformations raises an interesting point as to the possible diagnostic framing of the complex abnormality of the patient. We are grateful for the opportunity to discuss this case further and to give more clinical and radiological details, as suggested. Our patient has the bilateral syn- dactyly of the hands associated with forced flexion and soft tissue fusion. Radiographs showed five metacarpals, hypoplastic