

Genetic hypocalcaemia: a case of 22q deletion syndrome

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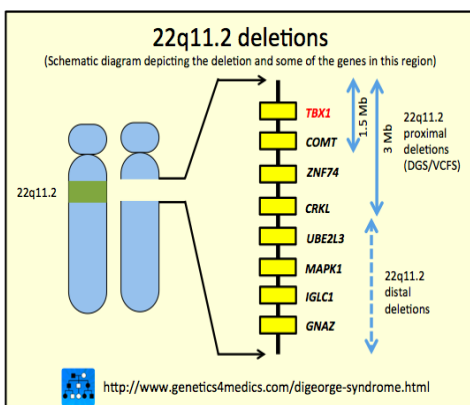
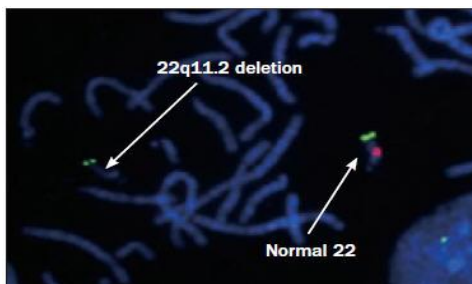
Introduction

22q11.2 deletion syndrome (velocardiofacial syndrome) is an autosomal dominant disorder that affects various organs including the parathyroid gland. Because of its incomplete penetrance, multi-system affection and variable clinical presentation, the diagnosis is often delayed for many years. Delayed diagnosis may have significant impact on morbidity and mortality. We present a patient with a long history of clinical features of a syndrome which was diagnosed after a delay of many years

Case

A 46-year-old man presented to the Emergency Department following a seizure. He had a low serum calcium level of 1.94mmol/L. After treatment he was lost to follow-up and re-presented four years later with a further seizure. His serum calcium level was 1.98mmol/L. He was prescribed calcium-vitamin D preparation and referred to the Endocrinology team. History taking elicited occasional seizures in childhood with no formal diagnosis of epilepsy. He had undergone a cleft palate repair as a child and had mild learning difficulties. On examination he had low-set ears and a scar indicating previous cleft palate repair. Further investigations revealed an inappropriately low serum parathyroid hormone level of 1.4pmol/L with normal vitamin D levels in the presence of hypocalcaemia. His clinical presentation led us to suspect a genetic mutation so he was referred to the medical geneticist for further evaluation. Micro-array studies confirmed a micro-deletion on the long arm of chromosome 22. Subsequent echocardiogram demonstrated a dilated aortic root awaiting cardiac MRI. An audiometry revealed sensory neuronal deafness.

Figure In this fluorescence in situ hybridization image, 2 green control probes are present, indicating chromosome 22. The missing red signal demonstrates the 22q deletion.



Individuals with a 22q deletion can have many possible features, ranging from the mild to the very serious

- Congenital heart disease
- Palatal abnormalities
- Learning difficulties
- Hypocalcaemia
- Significant feeding problems
- Renal anomalies
- Hearing loss (both conductive and sensorineural) (hearing loss with craniofacial syndromes)
- Laryngeal/tracheal/oesophageal anomalies
- Growth hormone deficiency
- Autoimmune disorders
- Immune disorders due to reduced T cell numbers
- Seizures (with or without hypocalcaemia)
- Skeletal abnormalities
- Psychiatric disorders

Conclusion

Hypocalcaemia in the young especially with phenotypic dimorphism should be addressed with increased diligence. We should have a low threshold for referral to medical genetics as they will require multidisciplinary team input and regular follow-up once a genetic diagnosis is established.