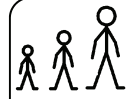


# Review Checklist

## 22q11 Deletion Syndrome



### CHILDREN (0—16 years)

Record **height, weight** and **head circumference measurements** in the **Child Health Record (Red Book)** and appropriate section of the **Personal Health Record (Blue Book)** if issued. Take **blood pressure** as soon as feasible.

#### HEART

Following diagnosis, all children should have a cardiology assessment which includes echocardiography.

If structural cardiac malformations are present, refer to Paediatric cardiologist.

#### PALATE

The palate should be examined. Ask about any history of feeding/swallowing difficulties and nasal regurgitation. Is there any evidence of nasal speech?

Refer to cleft palate team if symptomatic.

#### HYPOCALCAEMIA

Enquire about symptoms associated with hypocalcaemia which include tingling of the fingers and around the mouth. Use Chvostek sign to check for signs of tetany. Ask about muscle cramps, focal or generalised seizures, and apnoea in neonates. Check serum calcium level following diagnosis and at/during adolescent growth spurt

Refer to if low serum calcium levels. All patients on calcium supplements should have 2 yearly renal ultrasounds.

#### IMMUNE

Lymphocyte counts plus T and B cell subsets and immunoglobulin levels should be obtained following diagnosis. Repeat these tests at adolescence. Enquire about infection. Do not give live vaccines until these have been checked.

Refer to immunologist if test results are abnormal or if there are symptoms of autoimmune disorder.

#### DEVELOPMENT, EDUCATION & BEHAVIOUR

Is the child developing as expected? Formal developmental assessment should take place if the child is not reaching developmental milestones. Make enquiries about specific learning difficulties, behaviour and concentration span.

Refer to educational psychologist if not reaching developmental milestones or if there are significant learning or behaviour problems.

#### SPEECH & HEARING

Monitor language development and check for hypernasal speech. Enquire about hearing problems and infection. Children should have regular hearing tests. Continue to monitor language through secondary school from a speech and communication perspective

Refer to speech and language therapist for speech difficulties, audiology for hearing problems, and ENT for recurrent ear infections.

#### HYPOTHYROIDISM

Thyroid function tests are required after diagnosis and should be repeated if symptoms suggest hypothyroidism.

Refer to endocrinologist if thyroid function abnormal.

#### RENAL

A renal scan is required following diagnosis. Enquire about Urinary Tract Infections.

Refer to renal physician if recurrent UTIs or scan reveals a Structural malformation.

#### SKELETAL

X-Ray the spine if evidence of scoliosis and enquire about leg pain.

Refer to back surgeon if spinal anomaly and refer to orthotist if leg pain or flat feet.