

Extract from the Practical Guidelines
for Managing Patients with
22q11.2DS

Table II. Recommended assessments for 22q11.2 deletion syndrome *

| Assessment | At diagnosis | Infancy (0-12 months) | Preschool age (1-5 years) | School age (6-11 years) | Adolescence (12-18 years) | Adulthood (>18 years) |
|--|--------------|-----------------------|---------------------------|-------------------------|---------------------------|-----------------------|
| Ionized calcium, parathyroid hormone [†] | X | X | X | X | X | X |
| Thyrotropin (thyroid-stimulating hormone) [†] | X | | X | X | X | X |
| Complete blood cell count and differential (annual) | X | X | X | X | X | X |
| Immunologic evaluation ^z | X | X ^x | X ^x | | | |
| Ophthalmology | X | | X | | | |
| Evaluate palate ^f | X | X | X | | | |
| Audiology | X | X | X | | | X |
| Cervical spine (>age 4 years) | | | X ^{jj} | | | |
| Scoliosis examination | X | | X | | X | |
| Dental evaluation | | | X | X | X | X |
| Renal ultrasound | X | | | | | |
| Electrocardiogram | X | | | | | X |
| Echocardiogram | X | | | | | |
| Development ^{**} | X | X | X | | | |
| School performance | | | | X | X | |
| Socialization/functioning | X | X | X | X | X | X |
| Psychiatric/emotional/behavioral ^{††} | X | | X | X | X | X |
| Systems review | X | X | X | X | X | X |
| Deletion studies of parents | X | | | | | |
| Genetic counseling ^{zz} | X | | | | X | X |
| Gynecologic and contraceptive services | | | | | X | X |

*These recommendations are proposed as at year end 2010. Each U refers to a single assessment except as stated above and below. We have tended to err on the side of overinclusiveness. Local patterns of practice may vary.

[†]In infancy, test calcium levels every 3 to 6 months, then every 5 years through childhood, and every 1 to 2 years thereafter; thyroid studies annually. Check calcium preoperatively and postop-eratively and regularly in pregnancy.

^zIn addition to complete blood cell count with differential, in newborns: flow cytometry; and at age 9 to 12 months (before live vaccines): flow cytometry, immunoglobulins, T-cell function. Expert opinion is divided about the extent of needed immune work-up in the absence of clinical features.

^xEvaluate immune function before administering live vaccines (see ^z).

{In infancy, visualize palate and evaluate for feeding problems, nasal regurgitation, or both; in toddlers to adults, evaluate nasal speech quality.
^{jj}Cervical spine films to detect anomalies: anterior/posterior, lateral, extension, open mouth, skull base views. Expert opinion is divided about the advisability of routine radiography. Symptoms of cord compression are an indication for urgent neurological referral.

^{**}Motor and speech/language delays are common; rapid referral to early intervention for any delays can help to optimize outcomes.

^{††}Vigilance for changes in behavior, emotional state, and thinking, including hallucinations and delusions; in teens and adults, assessment would include at-risk behaviors (sexual activity, alcohol/ drug use, etc).

^{zz}See text for details.