## Recommended assessments for 22q11.2 deletion syndrome \*

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

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\*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. jjCervical 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 atrisk behaviors (sexual activity, alcohol/ drug use, etc).

zzSee text for details.