

 Shared health Soins communs Manitoba	22q11.2 FISH Testing Criteria	
	Approved by: Dr. A. Dawson	Effective Date: December 16, 2020

22q11.2 FISH testing may be ordered by clinical geneticists, pediatric cardiologists or pediatric immunologists as appropriate. Alternatively, microarray may be a more appropriate test and may be ordered by clinical geneticists only.

22q11.2 FISH TESTING CRITERIA

- A heart defect as listed below
 - Tetralogy of Fallot
 - Interrupted aortic arch
 - Truncus arteriosus
 - Pulmonary atresia
 - Double outlet right ventricle
 - Aberrant right subclavian artery
 - Vascular ring
 - Right sided aortic arch
- Parent, child or sibling with 22q11.2 deletion
- Positive NIPT for 22q11.2 deletion
- Cleft palate, submucous cleft palate, bifid uvula or velopharyngeal insufficiency
- DiGeorge phenotype (thymic aplasia, low calcium)
- Characteristic facial dysmorphism with bulbous tip of nose, cupped and over-folded helices of the ears and typical 'hooded' eyelids
- Schizophrenia with a history of cognitive impairment and physical features
- Positive SCID newborn screen (NBS)/immunodeficiency consistent with 22q11.2 deletion.
 - For all other immunological indications, please discuss with a Genomics laboratory genetic counsellor (204-787-4033) prior to ordering.