Proforma (22q11.2 microdeletion syndrome)

Name: Age: Sex: DOB:
Father:
Date (history taking): CR No.: OPD:
Occupation (parents): Education (parents):
Address:
Tel:
Email:

Pedigree

Referal Diagnosis:

Chief Complaints:

History:

Examination:
General:

Systemic:

Specific:

Column A	**Column B**	**Column C**
The presence of one of the following	*Two or more of the following core features*	*One core feature plus one of these associated features*
Conotruncal cardiac ano-maly such as Fallot's tetralogy, interrupted aortic arch, truncus arteriosus or major aorto-pulmonary collateral arteries	Characteristic facial abnormalities viz. broad bulbous nose, square shaped tip of nose, short filtrum, telecanthus, slant-ing eyes, low set ears, etc	Long slender fingers and hands
Parent of an affected child	Non-conotruncal congenital cardiac defect	Short stature
	Learning difficulties/deve-lopmental delay	Hypotonia
	Cleft palate, velopharyngeal insufficiency or swallowing difficulty	Renal abnormalities or Potter sequence
	Hypocalcaemia	Psychiatric (especially bipolar) disorders
	Immunodeficiency or thymic hypoplasia	Family history of congenital cardiac defects

Indications for FISH: Column A (one features); Columnn B (2 or more features); Column C (one column B + one column C features)