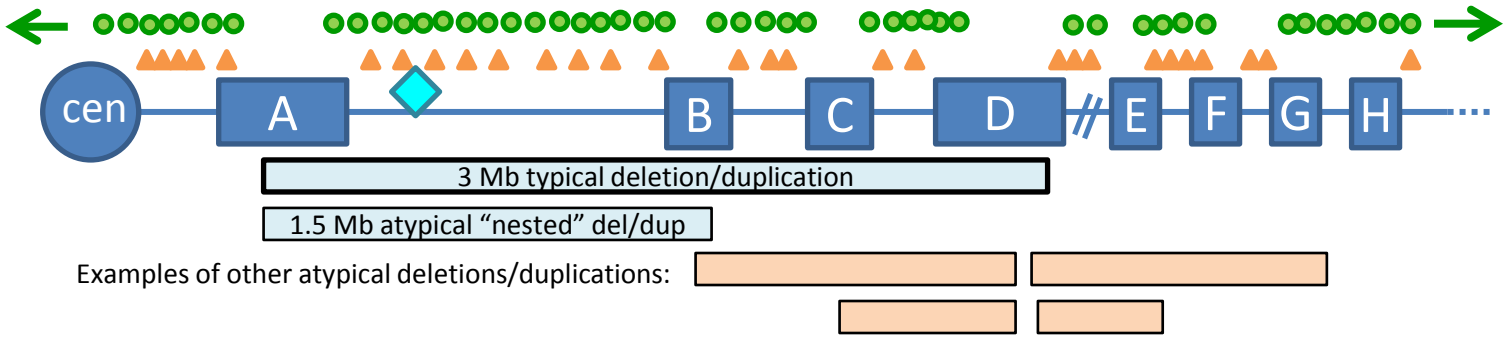
22q11.2 Deletion/Duplication Test Comparison Tool

Test Method:	MLPA	FISH	SNP Array
Detection rate of individuals with 22q11.2 deletion/duplication syndrome:	>98%	~95%	~100%
Detects typical deletions/duplications:	✓	✓	✓
Detects atypical deletions/duplications:	✓	Some	✓
Estimates size of deletion/duplication:	✓	X	✓
Detects other chromosomal disorders:	X	X	✓
Detects balanced rearrangements (i.e., in an unaffected parent, rare):	X	✓	X
Turn-around-time:	5-10 days	7-14 days	21 days
Blood sample collection container:	EDTA, ACD-A or -B	Sodium heparin	EDTA
Comparative charge:	$	$$	$$$$

22q11.2 Region Probe Coverage



Key:

- Single nucleotide polymorphism (SNP) array probes by whole genome assay
- Multiplex ligation-dependent probe amplification (MLPA) probes
- Low copy number repeats
- Fluorescence *in situ* hybridization (FISH) probe

References:

1) McDonald-McGinn DM, Zackai EH. Genetic counseling for the 22q11.2 deletion. Dev Disabil Res Rev. 2008;14:69–74, PMID: 18636638.

2) McDonald-McGinn DM, Emanuel BS, Zackai EH. 22q11.2 Deletion Syndrome. 1999 Sep 23 [Updated 2013 Feb 28]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1523/

3) Firth HV. 22q11.2 Duplication. 2009 Feb 17 [Updated 2013 Nov 21]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK3823/

4) Mikhail FM et al. The recurrent distal 22q11.2 microdeletions are often de novo and do not represent a single clinical entity: a proposed categorization system. Genet Med. 2014 Jan;16(1):92-100, PMID: 23765049.

5) Pinchefsky E, Laneuville L, Srour M. Distal 22q11.2 Microduplication: Case Report and Review of the Literature. Child Neurol Open. 2017 Nov 1;4:2329048X17737651, PMID: 29147671.

6) https://www.mlpa.com/WebForms/WebFormProductDetails.aspx?Tag=_tz2fAPIAupKyMjaDF-E-t9bmuxqlhe_Lgqfk8Hkjuss.&ProductOID=_0DLzOd5tLZ0.

Contact LabGC@seattlechildrens.org for more information

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