Prenatal Genomics Symposium

SurveyMonkey

1. Please provide other key issues that you/your institution wish to explore at the symposium.

	Which job role describes you best?				
	Clinical Geneticist	Laboratory Cytogeneticist/Molecular Geneticist	Clinical Genetic Counsellor	Obstetrician/Maternal Fetal Medicine Specialist	Response Count
	7 replies	6 replies	5 replies	1 reply	18
answered question	7	6	5	1	**18**
				skipped question	**22**

2. Which job role describes you best?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
Clinical Geneticist	**100.0% (16)**	21.4% (3)	0.0% (0)	0.0% (0)	**40.0% (16)**
Laboratory Cytogeneticist/Molecular Geneticist	18.8% (3)	**100.0% (14)**	0.0% (0)	0.0% (0)	35.0% (14)
Genetic technologist	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
Clinical Genetic Counsellor	0.0% (0)	0.0% (0)	**100.0% (11)**	0.0% (0)	27.5% (11)
Laboratory Genetic Counsellor	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
Obstetrician/Maternal Fetal Medicine Specialist	0.0% (0)	0.0% (0)	0.0% (0)	**100.0% (2)**	5.0% (2)
Bioethicist	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
Other (please specify)	0 replies	0 replies	0 replies	0 replies	0
answered question	16	14	11	2	**40**
				skipped question	**0**

3. Which institution do you work for ? (please answer to your comfort level -if you wish to maintain anonymity, please consider indicating Province only or may leave blank)

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Count**
	7 replies	9 replies	6 replies	0 replies	20
answered question	7	9	6	0	**20**
				skipped question	**20**

4. How often do you order prenatal chromosomal microarray or report out a prenatal microarray?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
Extremely often- every day	12.5% (2)	**21.4% (3)**	9.1% (1)	0.0% (0)	15.0% (6)
Very often-one or more a week	**50.0% (8)**	**21.4% (3)**	**45.5% (5)**	0.0% (0)	**37.5% (15)**
Moderately often-one or more times a month	18.8% (3)	14.3% (2)	18.2% (2)	**50.0% (1)**	17.5% (7)
Slightly often-less than once a month	18.8% (3)	**21.4% (3)**	18.2% (2)	0.0% (0)	17.5% (7)
Not at all	0.0% (0)	**21.4% (3)**	9.1% (1)	**50.0% (1)**	12.5% (5)
answered question	16	14	11	2	**40**
				skipped question	**0**

5. Do you think all pregnant women, irrespective of their a priori risk assessment, should be offered an invasive procedure, assuming adequate counselling regarding risks and benefits has been provided?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
yes	18.8% (3)	28.6% (4)	27.3% (3)	0.0% (0)	25.0% (10)
no	**68.8% (11)**	**57.1% (8)**	**54.5% (6)**	**100.0% (2)**	**62.5% (25)**
not sure	12.5% (2)	14.3% (2)	18.2% (2)	0.0% (0)	12.5% (5)
why?	11 replies	10 replies	5 replies	1 reply	27
answered question	16	14	11	2	**40**
				skipped question	**0**

6. At your institution, when do you currently offer prenatal chromosomal microarray after normal QFPCR/FISH aneuoploidy screen ? choose all that apply

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
We DO NOT offer prenatal chromosome microarray for any indication	0.0% (0)	7.7% (1)	0.0% (0)	0.0% (0)	2.6% (1)
When a parent is a carrier of a cryptic chromosomal rearrangement or had previous pregnancy/child with a microarray abnormality	**100.0% (16)**	**69.2% (9)**	90.9% (10)	**100.0% (1)**	**86.8% (33)**
When there is a fetal structural anomaly identified on ultrasound	87.5% (14)	**69.2% (9)**	**100.0% (11)**	**100.0% (1)**	**86.8% (33)**
Increased nuchal translucency	43.8% (7)	46.2% (6)	54.5% (6)	0.0% (0)	47.4% (18)
Cystic hygroma	62.5% (10)	53.8% (7)	81.8% (9)	0.0% (0)	68.4% (26)
Soft marker for aneuploidy	6.3% (1)	7.7% (1)	0.0% (0)	0.0% (0)	5.3% (2)
Positive screen for common aneuploidy	6.3% (1)	7.7% (1)	0.0% (0)	0.0% (0)	5.3% (2)
Advanced maternal age (greater than 35)	6.3% (1)	0.0% (0)	0.0% (0)	0.0% (0)	2.6% (1)
Maternal anxiety	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
ANY indication	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
I do not work in a prenatal setting	0.0% (0)	23.1% (3)	0.0% (0)	0.0% (0)	7.9% (3)

Other (please specify)	3 replies	6 replies	4 replies	1 reply	14
answered question	16	13	11	1	**38**
				skipped question	**2**

7. When do you think we should order a prenatal microarray (assume normal QFPCR/FISH for aneuoploidy)? Select all which apply.

		Which job role describes you best?				
		Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
Not for any indication	**yes**	10.0% (1)	27.3% (3)	0.0% (0)	0.0% (0)	
	no	**90.0% (9)**	**72.7% (8)**	**83.3% (5)**	**100.0% (2)**	
	not sure	0.0% (0)	0.0% (0)	16.7% (1)	0.0% (0)	
		10	11	6	2	27
When a parent is a carrier of a cryptic chromosomal rearrangement or had previous pregnancy/child with a microarray abnormality	**yes**	**93.8% (15)**	**85.7% (12)**	**90.9% (10)**	**100.0% (2)**	
	no	6.3% (1)	7.1% (1)	0.0% (0)	0.0% (0)	
	not sure	0.0% (0)	7.1% (1)	9.1% (1)	0.0% (0)	
		16	14	11	2	40
When there is a fetal structural anomaly identified on ultrasound	**yes**	**87.5% (14)**	**100.0% (14)**	**90.9% (10)**	**100.0% (2)**	
	no	6.3% (1)	0.0% (0)	0.0% (0)	0.0% (0)	
	not sure	6.3% (1)	0.0% (0)	9.1% (1)	0.0% (0)	

		16	14	11	2	40
Increased nuchal translucency	**yes**	**57.1% (8)**	**76.9% (10)**	**54.5% (6)**	**50.0% (1)**	
	no	14.3% (2)	15.4% (2)	27.3% (3)	0.0% (0)	
	not sure	28.6% (4)	7.7% (1)	18.2% (2)	**50.0% (1)**	
		14	13	11	2	38
Cystic hygroma	**yes**	**68.8% (11)**	**78.6% (11)**	**90.9% (10)**	**100.0% (2)**	
	no	0.0% (0)	7.1% (1)	0.0% (0)	0.0% (0)	
	not sure	31.3% (5)	14.3% (2)	9.1% (1)	0.0% (0)	
		16	14	11	2	40
Soft marker for aneuploidy	**yes**	0.0% (0)	**46.2% (6)**	10.0% (1)	0.0% (0)	
	no	**72.7% (8)**	**46.2% (6)**	**70.0% (7)**	0.0% (0)	
	not sure	27.3% (3)	7.7% (1)	20.0% (2)	**100.0% (2)**	
		11	13	10	2	34
Positive screen for common aneuploidy	**yes**	8.3% (1)	**46.2% (6)**	10.0% (1)	0.0% (0)	
	no	**75.0% (9)**	**46.2% (6)**	**80.0% (8)**	33.3% (1)	
	not sure	16.7% (2)	7.7% (1)	10.0% (1)	**66.7% (2)**	
		12	13	10	3	35
Advanced maternal age (greater than 35)	**yes**	8.3% (1)	23.1% (3)	10.0% (1)	0.0% (0)	

	no	**66.7% (8)**	**53.8% (7)**	**80.0% (8)**	0.0% (0)	
	not sure	25.0% (3)	23.1% (3)	10.0% (1)	**100.0% (2)**	
		12	13	10	2	35
Maternal anxiety	**yes**	8.3% (1)	23.1% (3)	0.0% (0)	0.0% (0)	
	no	**66.7% (8)**	**69.2% (9)**	**80.0% (8)**	**50.0% (1)**	
	not sure	25.0% (3)	7.7% (1)	20.0% (2)	**50.0% (1)**	
		12	13	10	2	35
ANY indication	**yes**	0.0% (0)	23.1% (3)	0.0% (0)	0.0% (0)	
	no	**88.9% (8)**	**69.2% (9)**	**77.8% (7)**	**100.0% (2)**	
	not sure	11.1% (1)	7.7% (1)	22.2% (2)	0.0% (0)	
		9	13	9	2	31
Other (please specify)		8 replies	4 replies	3 replies	1 reply	16
answered question		16	14	11	2	**40**
					skipped question	**0**

8. Under which circumstances do you think a reflex prenatal karyotype is warranted?

		Which job role describes you best?				
		Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
all indications when microarray with SNP is normal	**yes**	13.3% (2)	0.0% (0)	30.0% (3)	0.0% (0)	
	no	**80.0% (12)**	**100.0% (13)**	**50.0% (5)**	**50.0% (1)**	
	not sure	6.7% (1)	0.0% (0)	20.0% (2)	**50.0% (1)**	
		15	13	10	2	37
parental balanced translocation when microarray with SNP is normal	**yes**	33.3% (5)	**57.1% (8)**	**60.0% (6)**	**50.0% (1)**	
	no	**53.3% (8)**	42.9% (6)	30.0% (3)	0.0% (0)	
	not sure	13.3% (2)	0.0% (0)	10.0% (1)	**50.0% (1)**	
		15	14	10	2	38
unbalanced fetal microarray revealing a deletion/duplication	**yes**	**68.8% (11)**	**41.7% (5)**	**54.5% (6)**	**50.0% (1)**	
	no	18.8% (3)	**41.7% (5)**	45.5% (5)	0.0% (0)	
	not sure	12.5% (2)	16.7% (2)	0.0% (0)	**50.0% (1)**	
		16	12	11	2	38
abnormal QFPCR indicating common anueoploidy (note some centres considering parental karyotypes to	**yes**	**87.5% (14)**	**76.9% (10)**	**90.9% (10)**	0.0% (0)	
	no	6.3% (1)	23.1% (3)	9.1% (1)	**50.0% (1)**	

avoid need for prenatal cell cultures in such cases)	**not sure**	6.3% (1)	0.0% (0)	0.0% (0)	**50.0% (1)**	
		16	13	11	2	39
Other (please specify)		3 replies	4 replies	1 reply	0 replies	8
answered question		16	14	11	2	**40**
					skipped question	**0**

9. If prenatal microarray is to be done, should there be a consent form? And if so, what key issues/points should be addressed?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
no	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
yes-please comment on key points below	**87.5% (14)**	**100.0% (14)**	**81.8% (9)**	**100.0% (2)**	**90.0% (36)**
not sure	12.5% (2)	0.0% (0)	18.2% (2)	0.0% (0)	10.0% (4)
why? and if yes, key points...	14 replies	13 replies	11 replies	2 replies	40
answered question	16	14	11	2	**40**
				skipped question	**0**

10. Should patients have the opportunity, through the consent process, to select what kind of data, if any, they would like withheld from the report? If yes, check all which apply.

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
no	0.0% (0)	14.3% (2)	0.0% (0)	0.0% (0)	5.0% (2)
yes-can elect to withhold Variants of Unknown Significance (VOUS)	68.8% (11)	50.0% (7)	63.6% (7)	**100.0% (2)**	60.0% (24)
yes-can elect to withhold findings associated only with adult onset conditions for which their is no treatment	**100.0% (16)**	**71.4% (10)**	**72.7% (8)**	**100.0% (2)**	**82.5% (33)**
yes- can elect to withhold findings only revealing an autosomal recessive carrier state	56.3% (9)	35.7% (5)	36.4% (4)	**100.0% (2)**	50.0% (20)
yes- can elect to withhold findings only disclosing the possibility of non-paternity	75.0% (12)	35.7% (5)	36.4% (4)	**100.0% (2)**	52.5% (21)
yes- can elect to withhold findings only disclosing the possibility of consangunity	50.0% (8)	28.6% (4)	18.2% (2)	**100.0% (2)**	37.5% (15)
yes- can elect to withhold findings only related to uniparental disomy	12.5% (2)	14.3% (2)	18.2% (2)	**100.0% (2)**	20.0% (8)
yes-other: please comment	6.3% (1)	0.0% (0)	0.0% (0)	0.0% (0)	2.5% (1)
not sure	18.8% (3)	14.3% (2)	27.3% (3)	0.0% (0)	20.0% (8)
Why?	5 replies	6 replies	5 replies	1 reply	17

answered question	16	14	11	2	**40**
				skipped question	**0**

11. Regarding Variants of Unknown Significance (VOUS) and Copy Number Variants (CNV), un which circumstances should they be reported? NOTE: IF the format of this question does not s how you would wish to respond, please feel free to comment using the text box below.

		Which job role describes you best?				
		Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Re T**
benign CNV	**always**	7.1% (1)	7.1% (1)	20.0% (2)	0.0% (0)	
	only if patient consents to knowing this information	14.3% (2)	0.0% (0)	20.0% (2)	0.0% (0)	
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	
	only if a post-natal concern is raised	7.1% (1)	0.0% (0)	0.0% (0)	0.0% (0)	
	not sure	0.0% (0)	0.0% (0)	10.0% (1)	**100.0% (2)**	
	never	**71.4% (10)**	**92.9% (13)**	**50.0% (5)**	0.0% (0)	
		14	14	10	2	
pathogenic CNV (within targeted	**always**	**85.7% (12)**	**86.7% (13)**	**66.7% (8)**	0.0% (0)	

region or based on size/genetic content predicted to be pathogenic)	**only if patient consents to knowing this information**	0.0% (0)	0.0% (0)	16.7% (2)	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	6.7% (1)	8.3% (1)	0.0% (0)
	only if a post-natal concern is raised	14.3% (2)	6.7% (1)	8.3% (1)	0.0% (0)
	not sure	0.0% (0)	0.0% (0)	0.0% (0)	**100.0% (2)**
	never	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
		14	15	12	2
likely pathogenic CNVs (based on size/gene content predicted to have ability to be pathogenic)	**always**	**78.6% (11)**	**60.0% (9)**	**80.0% (8)**	0.0% (0)
	only if patient consents to knowing this information	7.1% (1)	26.7% (4)	10.0% (1)	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	6.7% (1)	10.0% (1)	0.0% (0)
	only if a post-natal concern is raised	14.3% (2)	6.7% (1)	0.0% (0)	0.0% (0)

	not sure	0.0% (0)	0.0% (0)	0.0% (0)	**100.0% (2)**
	never	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
		14	15	10	2
VOUS-with genes not known to be associated with a genetic disorder	**always**	7.1% (1)	0.0% (0)	23.1% (3)	0.0% (0)
	only if patient consents to knowing this information	**42.9% (6)**	35.7% (5)	**38.5% (5)**	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	7.1% (1)	7.1% (1)	15.4% (2)	0.0% (0)
	only if a post-natal concern is raised	7.1% (1)	7.1% (1)	7.7% (1)	0.0% (0)
	not sure	28.6% (4)	7.1% (1)	7.7% (1)	**100.0% (2)**
	never	7.1% (1)	**42.9% (6)**	7.7% (1)	0.0% (0)
		14	14	13	2
VOUS-with genes associated with a disorder but not known if expression is affected e.g. partial gene duplication or duplication found but disorder associated with deletion only	**always**	**28.6% (4)**	6.7% (1)	20.0% (3)	0.0% (0)
	only if patient consents to knowing this information	**28.6% (4)**	**33.3% (5)**	**40.0% (6)**	0.0% (0)
	only if				

	there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	6.7% (1)	13.3% (2)	0.0% (0)
	only if a post-natal concern is raised	7.1% (1)	6.7% (1)	13.3% (2)	0.0% (0)
	not sure	21.4% (3)	26.7% (4)	13.3% (2)	**100.0% (2)**
	never	14.3% (2)	20.0% (3)	0.0% (0)	0.0% (0)
		14	15	15	2
CNVs known to be only associated with a predisposition to neurodevelopmental disorders (eg autism)	**always**	25.0% (4)	26.7% (4)	33.3% (3)	0.0% (0)
	only if patient consents to knowing this information	**56.3% (9)**	**40.0% (6)**	**66.7% (6)**	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
	only if a post-natal concern is raised	0.0% (0)	6.7% (1)	0.0% (0)	0.0% (0)
	not sure	18.8% (3)	13.3% (2)	0.0% (0)	**100.0% (2)**
	never	0.0% (0)	13.3% (2)	0.0% (0)	0.0% (0)
		16	15	9	2

CNVs known to be only associated with an adult onset condition (no current treatment exists)	**always**	7.1% (1)	7.7% (1)	8.3% (1)	0.0% (0)
	only if patient consents to knowing this information	**57.1% (8)**	**46.2% (6)**	**66.7% (8)**	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
	only if a post-natal concern is raised	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
	not sure	14.3% (2)	30.8% (4)	8.3% (1)	**100.0% (2)**
	never	21.4% (3)	15.4% (2)	16.7% (2)	0.0% (0)
		14	13	12	2
CNVs revealing an autosomal recessive condition	**always**	26.7% (4)	13.3% (2)	**38.5% (5)**	0.0% (0)
	only if patient consents to knowing this information	**40.0% (6)**	**33.3% (5)**	30.8% (4)	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	13.3% (2)	7.7% (1)	0.0% (0)
	only if a post-natal	0.0%	6.7%	7.7%	0.0%

	concern is raised	(0)	(1)	(1)	(0)
	not sure	20.0% (3)	13.3% (2)	7.7% (1)	**100.0% (2)**
	never	13.3% (2)	20.0% (3)	7.7% (1)	0.0% (0)
		15	15	13	2
CNVs with only involving genes on the American College of Medical Genetics incident findings list	**always**	30.8% (4)	23.1% (3)	18.2% (2)	0.0% (0)
	only if patient consents to knowing this information	23.1% (3)	23.1% (3)	**45.5% (5)**	0.0% (0)
	only if there is a structural anomaly (hard finding) on ultrasound	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
	only if a post-natal concern is raised	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
	not sure	**38.5% (5)**	**46.2% (6)**	36.4% (4)	**100.0% (2)**
	never	7.7% (1)	7.7% (1)	0.0% (0)	0.0% (0)
		13	13	11	2
Please comment		6 replies	3 replies	2 replies	0 replies
answered question		14	14	10	2
					skipped question

12. Should a Canadian guideline for prenatal reporting of Variants of Unknown Significance be developed?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Totals**
yes	**81.3% (13)**	**92.9% (13)**	**81.8% (9)**	**100.0% (2)**	**85.0% (34)**
no	6.3% (1)	7.1% (1)	0.0% (0)	0.0% (0)	5.0% (2)
not sure	12.5% (2)	0.0% (0)	18.2% (2)	0.0% (0)	10.0% (4)
Why?	9 replies	8 replies	5 replies	1 reply	23
answered question	16	14	11	2	**40**
				skipped question	**0**

13. Any other issues you would like to have addressed? Comments?

	Which job role describes you best?				
	Clinical Geneticist	**Laboratory Cytogeneticist/Molecular Geneticist**	**Clinical Genetic Counsellor**	**Obstetrician/Maternal Fetal Medicine Specialist**	**Response Count**
	3 replies	4 replies	1 reply	0 replies	8
answered question	3	4	1	0	**8**
				skipped question	**32**