Assessing the Economics of Genomic Medicine: A Workshop

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DISCLOSURE

- The Department of Molecular and Human Genetics at Baylor College of Medicine (BCM) offers extensive genetic laboratory testing, and the Department derives revenue from this activity.
- NY Times Saturday July 14, 2012

Conflict Potential Seen in Genetic Counselors

- Genetic testing raises some vexing ethical questions, like whether it will cause unnecessary anxiety or lead to more medical procedures, including abortions.

•NY Times Saturday July 14, 2012

Conflict Potential Seen in Genetic Counselors

- **Now, as the number of tests and the money to be made from them are exploding, another question is being asked by professionals in the field themselves. Is it ethical for genetic counselors, who advise patients on whether to undergo testing, to be paid by the companies that perform the tests?**

•NY Times Saturday July 14, 2012

Futurist perspective

All individuals may already know the WGS of their parents.

Most fetuses may have WGS determined in the first trimester using non-invasive prenatal diagnosis.

GROUP I RISKS

- Mutations that she might have that could impact her own health; this could include BRCA1/2 mutation, HNPCC cancer, familial hypercholesterolemia, and ApoE

GROUP II RISKS

- Genetic risk to her offspring based on inheritied mutations; this would include anything identified in Group 1 above and carrier testing for autosomal recessive and some X-linked disorders

GROUP III RISKS

- Gentic risks related to new mutations such as trisomies, point mutations such as neurexin 1 and achondroplasia, and deletion/duplications such as DiGeorge syndrome.

TWO CATEGORIES OF SEVERITY

- Phenotypes where a person cannot achieve normal employment or live fully independently. Affected individuals don't advocate. Many families chose abortion.
  - Tay Sachs, trisomy 18, Down syndrome,
- Phenotypes with milder severity. Families rarely chose abortion. Preimplantation genetic diagnosis (PGD) is an attractive option.
  - BRCA1, hereditary deafness, achondroplasia

Futurist perspective

- 1) What is the theoretical economic rationale for developing targeted versus WGS approaches in your clinical scenario?

Futurist perspective

- WGS more expensive initially but still may be cost effective if very expensive phenotypes are avoided.
- Phenotypes where a person can never be employed or live independently.
- From a multigenerational perspective, WGS is far more cost effective.
- Pre-existing data.

Futurist perspective

- 2) What is known and not known about the type of variations (including their potential clinical utility) that might be found in each of these scenarios? How is this likely to change? How will this effect models for implementation?

Futurist perspective

- We must identify the causative mutations for all individuals on the planet with serious Mendelian disorders.
- Implementation of prevention should focus on preimplantation genetic diagnosis (PGD) for inherited mutations.

Futurist perspective

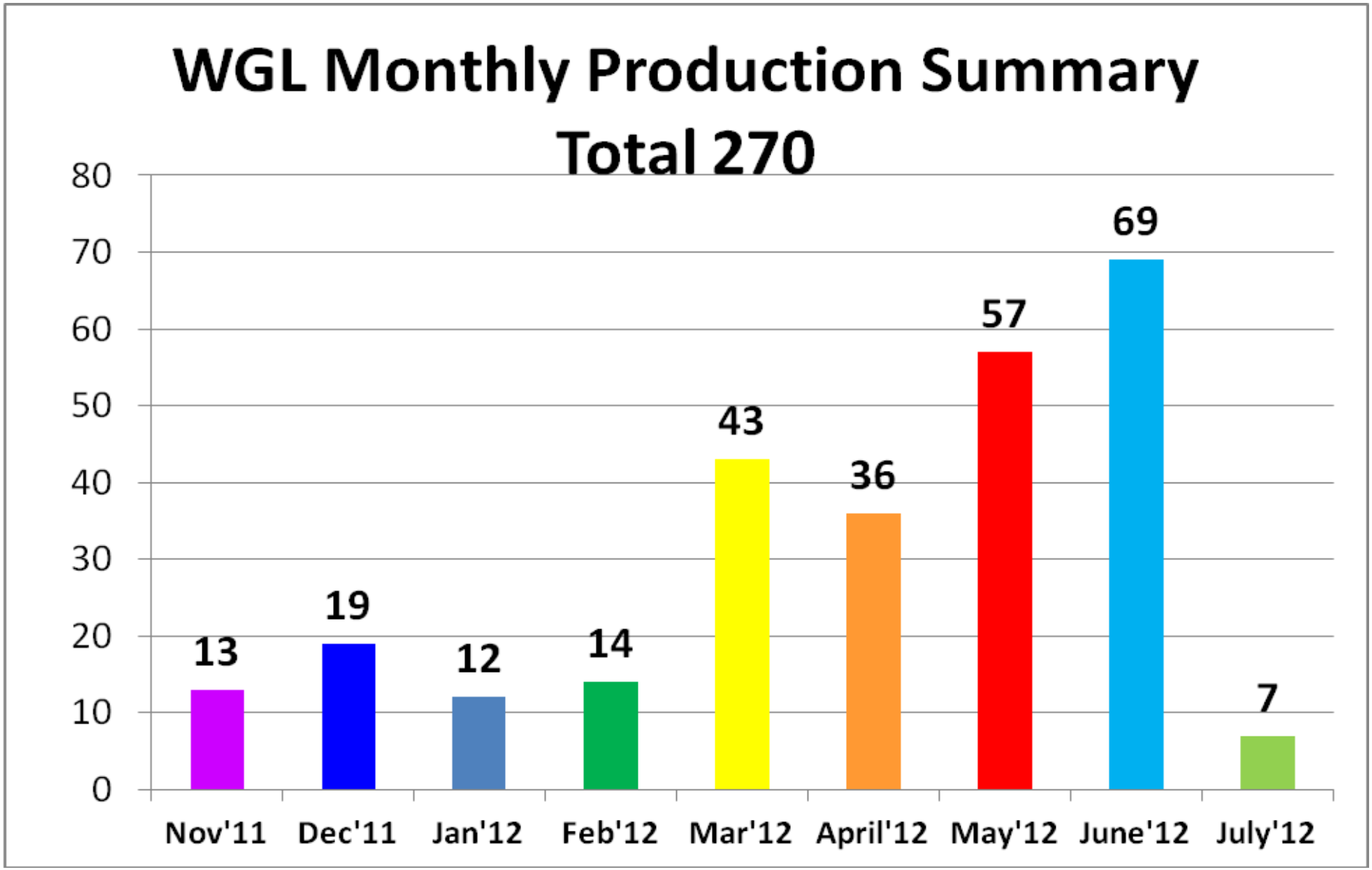
- ***De novo*** **mutations can only be detected by intra-partum testing.**

Futurist perspective

- 3) How should “secondary findings” that might arise under each of the clinical scenarios be dealt with? What is known about health care provider and patient behaviors when confronted with such incidental findings?

Futurist perspective

- Secondary finding must be dealt with on a case by case basis.
- I believe that more information is almost always better. We don’t offer limited physical exams.
- The behavior of providers must be regulated.
- Most patients can be counseled through informed decision making.


WGL Monthly Production Summary
Total 270
80
70
60
50
40
30
20
10
0
13
19
12
14
43
36
57
69
7
Nov'11
Dec'11
Jan'12
Feb'12
Mar'12
April'12
May'12
June'12
July'12

	Gene	Mutation	Associated Phenotype	Inheritance
1	ARID1B	nonsense mutation	Mental retardation, Coffin Siris	AD
2	CASK	two-nucleotide duplication (mosaic)	Mental retardation	X-linked
3	CBL	splicing mutation	Noonan syndrome-like disorder	AD
4	MECP2	one-nucleotide deletion	Rett ; Encephalopathy, neonatal, severe	X-linked
5	NDUFV1	homozygous missense, previously reported as deleterious mutation	Mitochondrial complex I deficiency	AR
6	PTPN11	missense, common deleterious	Noonan syndrome, LEOPARD	AD
7	RBM10	splicing mutation	TARP syndrome	X-linked
8	SACS	1) nonsense, 2) two-nucleotide del	Spastic ataxia, Charlevoix-Saguenay	AR
9	SACS	Homozygous two-nucleotide del	Spastic ataxia, Charlevoix-Saguenay	AR

END

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http://www.bcm.edu/geneticlabs/

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