# NEWSLETTER

This newsletter centers on reproductive topics with a genetics focus. If there is an organization or upcoming webinar you'd like me to include in a future newsletter, please feel free to reach out at genetics@modernreproduction.org.

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## Webinars

### LARGE LANGUAGE MODEL AI CHATBOTS REQUIRE APPROVAL AS MEDICAL DEVICES

October 17, 2023 @ **12:00** pm ET

Register

### NAVIGATING STATE BIOMARKER LEGISLATION: IMPLICATIONS & IMPLEMENTATION

October 17, 2023 @ **2:00** pm ET

Register

# NEWSLETTER

## *The little lit review*

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### Pregnant people's views and knowledge on prenatal screening for fetal trisomy in the absence of a national screening program

Kristin Kelly| Sara Leitao| Sarah Meaney| Keelin O'Donoghue

332 pregnant women in Ireland were surveyed in order to better understand their knowledge on screening options available. 227 women had another child. About 63% of participants had not heard of NIPT, though 92% would proceed with NIPT if offered. The results of the study could be influential to the desire for a screening program in Ireland. Another source of value are areas to improve for patient education and awareness as well as understanding patient motivations for testing. I do wonder, however, how much this concept of healthy is personal or societal, and what does healthy mean to a person.

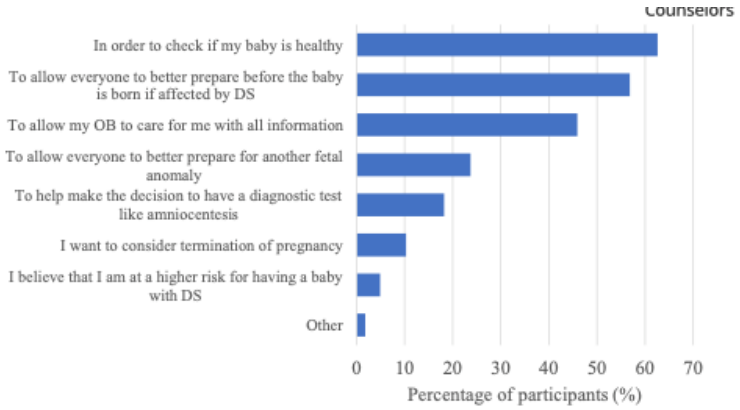


FIGURE 1 Reasons for which participants would accept prenatal screening tests. DS, Down Syndrome; OB, Obstetrician.

TABLE 2 Participants' knowledge on screening tests.

| | Correct *n* (%) | Incorrect *n* (%) | Unsure *n* (%) |
|---|---|---|---|
| Current cost of prenatal screening test (*n* = *270*) | 101 (37.1) | 171 (62.9) | |
| Screening can detect all babies with disease/disability (*n* = *321*) | 200 (62.3) | 52 (16.2) | 69 (21.5) |
| Can screening detect all babies with Down syndrome? (*n* = *328*) | 201 (61.3) | 50 (15.2) | 77 (23.5) |
| Interpretation of low-risk screening test result for DS (*n* = *326*) | 251 (77.0) | 31 (9.5) | 44 (13.5) |
| Interpretation of high-risk screening test result for DS (*n* = *323*) | 245 (75.9) | 31 (9.6) | 47 (14.6) |
| Current rate of Down syndrome in Ireland (*n* = 324) | 50 (15.4) | 72 (22.2) | 202 (62.3) |
| Are most Down syndrome babies born to women >35 years old? (*n* = *323*) | 124 (38.4) | 107 (33.1) | 92 (28.5) |
| "Screening for anomalies" understanding (*n* = 330)[a] | | | 21 (6.4) |
| Anatomy scan | 257 (77.9) | 73 (22.1) | |
| Blood tests to look at levels in the mother's blood | 46 (13.9) | 284 (86.1) | |
| Measuring fluid on back of baby's neck | 107 (32.4) | 223 (67.6) | |
| Ultrasound scan to look for signs of Down syndrome | 143 (43.3) | 187 (56.7) | |
| Blood tests to look at the baby's DNA for risk of Down syndrome | 139 (42.1) | 191 (57.9) | |

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# NEWSLETTER

## *The little lit review*

### An overview of reproductive carrier screening panels for autosomal recessive and/or X-linked conditions: How much do we know?

Tianjiao Wang | Paul Scuffham | Joshua Byrnes | Martin B. Delatycki | Martin Downes

The authors explored the various carrier screening panels available to the public from the US, Australia, UK, Netherlands, Spain, Germany, and Italy. 22 panels were identified through google search, other literature searches, and email. The size of the panel range from 44 to 2054 genes - most were >100 and <1000. 15 genes were found within each panel.

There is debate as to which genes to include on a panel. From the commercial perspective, more may be perceived as a better value, yet more may not translate to higher clinical utility. ACMG attempts a framework, focusing on severity and carrier frequency.

Determining which genes based on severity is highly subjective and may even be clinically incorrect given continually new updates to genotype-phenotype correlations and treatment options. Basing the decision on carrier frequency may not be a great metric as well as noted by several other papers cited in the paper.

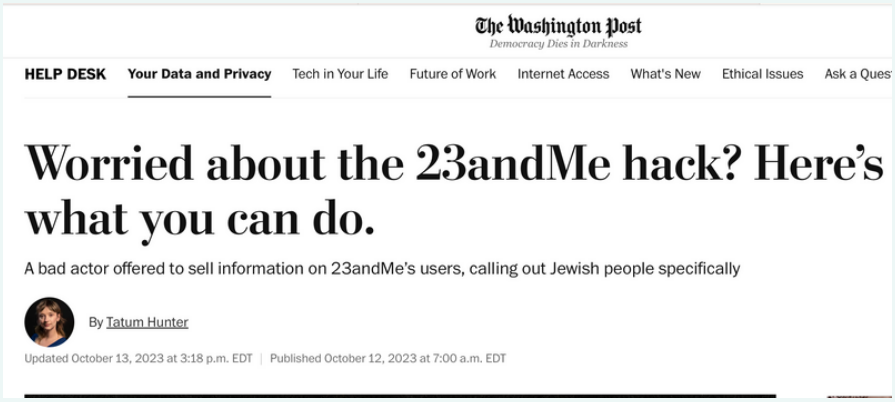
The authors do not offer a solution of how to select which genes to include on a carrier screening panel, yet they raise that by not having stringent criteria, market competition can continue. I've always thought streamlining and consistency is the way to go with healthcare, but it's a fair thought that we may never accomplish this in the carrier screening world and instead should embrace the commercial approach.

TABLE 3 The 15 genes selected by 22 identified providers.

| Gene | OMIM phenotype | Carrier frequency reported by invitae (1 in) | Carrier frequency reported by Monash IVF (1 in) | Severity reported by CentoGene |
|---|---|---|---|---|
| *HBB* | Thalassemias, beta-, 613985 | 49 | 158 | Mild, moderate or severe |
| *PEX1* | Peroxisome biogenesis disorder 1A (Zellweger), 214100 | 144 | 147 | Mild, moderate or severe |
| *PEX7* | Chondrodysplasia punctata, rhizomelic, type 1, 215100 | 157 | 158 | Mild, moderate or severe |
| *ASPA* | Canavan disease, 271900 | 159 | 300 | Moderate or severe |
| *PMM2* | Congenital disorder of glycosylation, type Ia, 212065 | 190 | >500 | Severe |
| *PPT1* | Ceroid lipofuscinosis, neuronal, 1, 256730 | 199 | 368 | Severe |
| *HEXA* | Tay-Sachs disease, 272800 | 250 | 300 | Moderate or severe |
| *SMPD1* | Niemann-Pick disease, type A, 257200 | 250 | 250 | Moderate or severe |
| *FANCC* | Fanconi anemia, complementation group C, 227645 | 417 | 535 | Severe |
| *NBN* | Nijmegen breakage syndrome, 251260 | ≥500 | 158 | Severe |
| *ALDH3A2* | Sjogren-Larsson syndrome, 270200 | ≥500 | 250 | Severe |
| *BLM* | Bloom syndrome, 210900 | ≥500 | 800 | Severe |
| *CLN5* | Ceroid lipofuscinosis, neuronal, 5, 256731 | ≥500 | >500 | Severe |
| *ELP1/IKBKAP* | Dysautonomia, familial, 223900 | ≥500 | 300 | Severe |
| *MCOLN1* | Mucolipidosis IV, 252650 | ≥500 | 300 | Moderate or severe |

# NEWSLETTER

## Community Content:




This week's community content spotlight is more on recent events than a resource available. Why? Because while we discuss potential issues within genetics, we should be aware of the times that those concerns come to fruition. Patients may be particularly sensitive to a lab obtaining their data. It's helpful to know the lab's policies on data protection but also retainment. Some labs may retain DNA information for internal developmental processes.

## Modern Reproduction Content:

These videos have been nothing but fun :D