

Inconsistencies in Carrier Screening Guidelines Result in Inconsistencies in Equitable Fetal Risk Detection



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Background

- Carrier screening for recessive disease risk is an important component of reproductive risk assessment for pregnant and prospectively pregnant patients
- Discrepancies in screening guidelines result in a wide variety of screening strategies that providers and patients may pursue
 - ACOG currently emphasizes provider consistency over a specific panel and recommends universal screening for only 2 conditions for all pregnancies
 - cystic fibrosis (CF)
 - spinal muscular atrophy (SMA)
 - ACMG recommends universal screening for 113 conditions
- We examined carrier screening panel ordering patterns in a commercial laboratory by provider type

Methods

- We retrospectively analyzed orders placed for carrier screening from a commercial laboratory (Foresight; Myriad Genetics, Inc.) between 07/2024-06/2025
- Orders were stratified by type of medical provider placing the order, with each stratum having >1000 orders:
 - OB/Gyn
 - MFM
 - Internal Medicine
 - Genetics
- IVF and unspecified provider types were excluded from this analysis
- Panel classification details are described in **Table 1**
- Panels with fewer than 40 orders associated were excluded from analysis

Table 1. Panel Classification

Panel	Description
Fundamental Panel (F)	2-3 conditions always including CF and SMA
Fundamental Plus (F+)	14 conditions
Universal (U)	176 conditions
Universal Plus (U+)	267 conditions; includes the majority of the ACMG recommended
Custom Panel	Panels not fitting above criteria; can be comprised of any combination of available genes and can span 1-266 conditions. <small>*26% of these panels were reflex screening of reproductive partners of carriers.</small>

Results

- Ob/Gyn accounted for the majority of carrier screening order (**Figure 1**)
- Panel ordering patterns varied by provider type (**Table 2 & Figure 2**)
- Although OB/Gyn providers accounted for the majority of carrier screening orders, they order larger U and U+ panels only 31.7% of the time. The majority of their orders (53%) placed are for the smaller F and F+ panels
- Genetics providers in contrast ordered larger U and U+ panels 74.2% of the time
- MFM providers order larger U and U+ panels 52% of the time
- Internal medicine providers, who represent 0.7% of orders placed, order larger panels 82% of the time

Figure 1. Carrier Screening Ordering by Provider Type

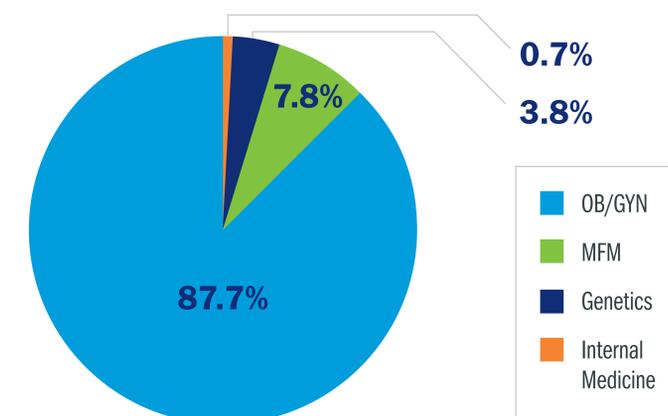
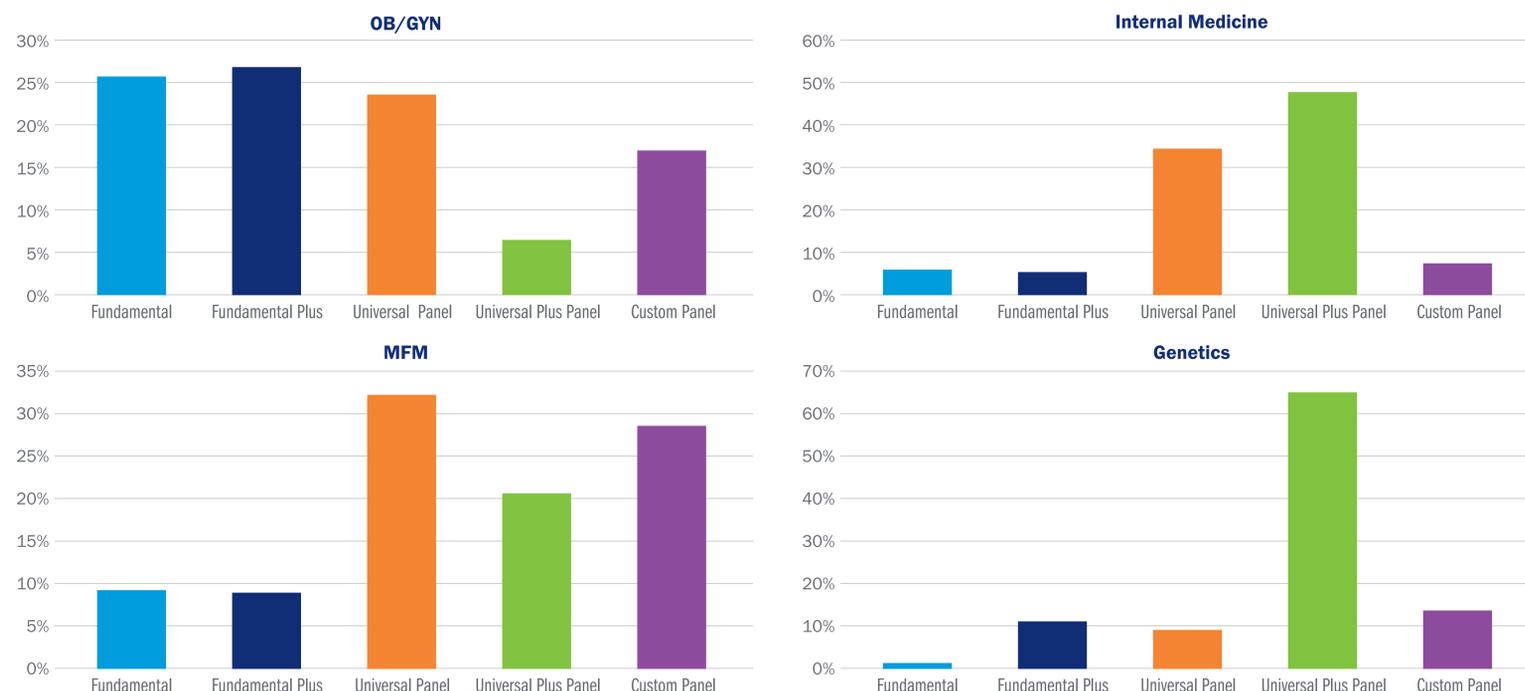


Table 2. Panel Ordering Patterns (%)

Provider Type	Fundamental (%)	Fundamental + (%)	Universal (%)	Universal + (%)	Custom (%)
OB/Gyn	26	27	25	6.7	17
MFM	9.2	11	32	20	28
Genetics	1	11	9.2	65	14
Internal Medicine	5.5	5.2	34	48	7

Figure 2. Panel Ordering by Provider Type



Conclusions

- OB/Gyn, the specialty that cares for the majority of pregnant patients in the US only order larger carrier screening panels most aligned with ACMG goals of carrier screening 31.7% of the time.
- Only patients with qualifying risk factors are referred to or co-managed by specialists such as MFM or genetics, suggesting that despite the

ACMG guidance, many patients are not screened sufficiently to achieve the goal of equitable and effective screening for a diverse population

- Unified guidelines across medical societies that provide guidance for pregnancy care could potentially help bridge the current gaps in care