

# Analyzing Pediatrician Attitudes and Understandings of BabySeq Genome Sequencing Reports

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## Results

### Demographics

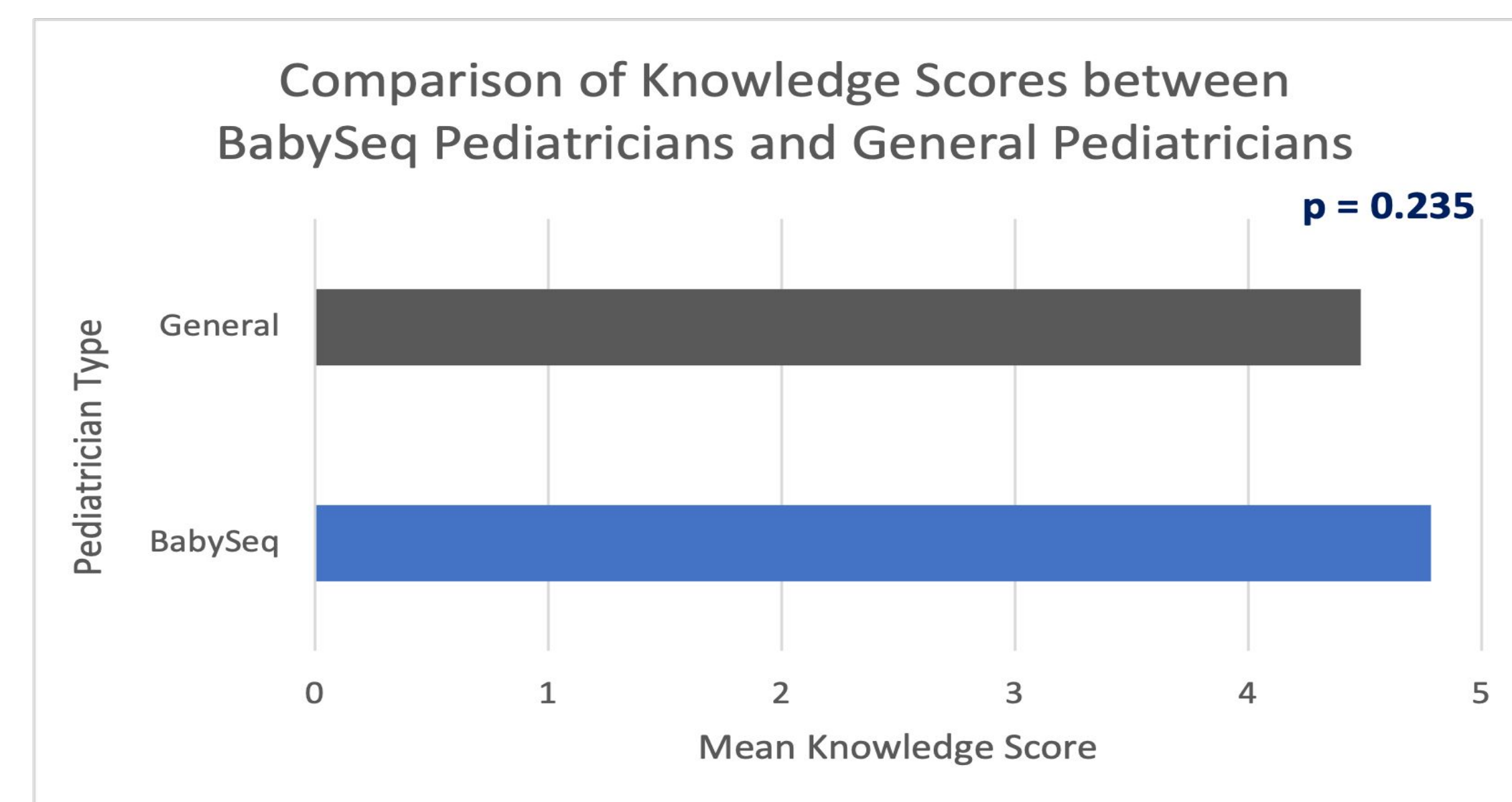
20 pediatricians completed survey

Female	80%
White	65%
31-40 Years Old	40%
No BabySeq Training	70%

### Pediatrician Knowledge of GS Reports

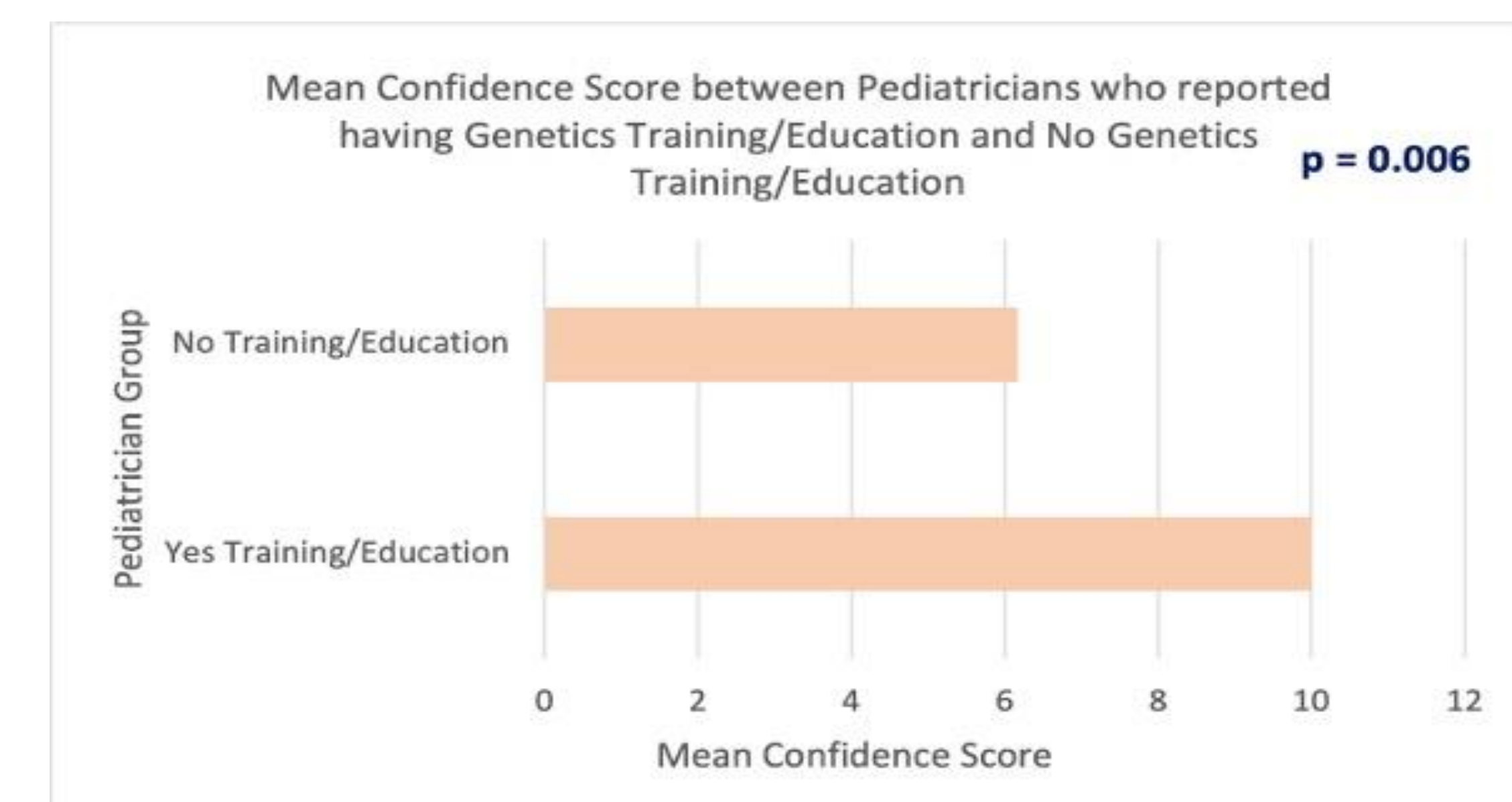
Concepts Performed the Highest	Concepts Performed the Lowest
Identification of what conditions the patient is at risk for versus not	Carrier testing utility and implications for family members
Recommendations for when referrals are necessary	Management of low penetrance findings

### Comparison of Knowledge Scores



- BabySeq Pediatricians had a slightly higher mean knowledge score than General Pediatricians (this difference was not statistically significant)

### Comparison of Confidence Scores



- Pediatricians who reported having any genetics training/education, including outside of BabySeq, had significantly more confidence in reviewing, ordering, communicating, and managing genetic test results

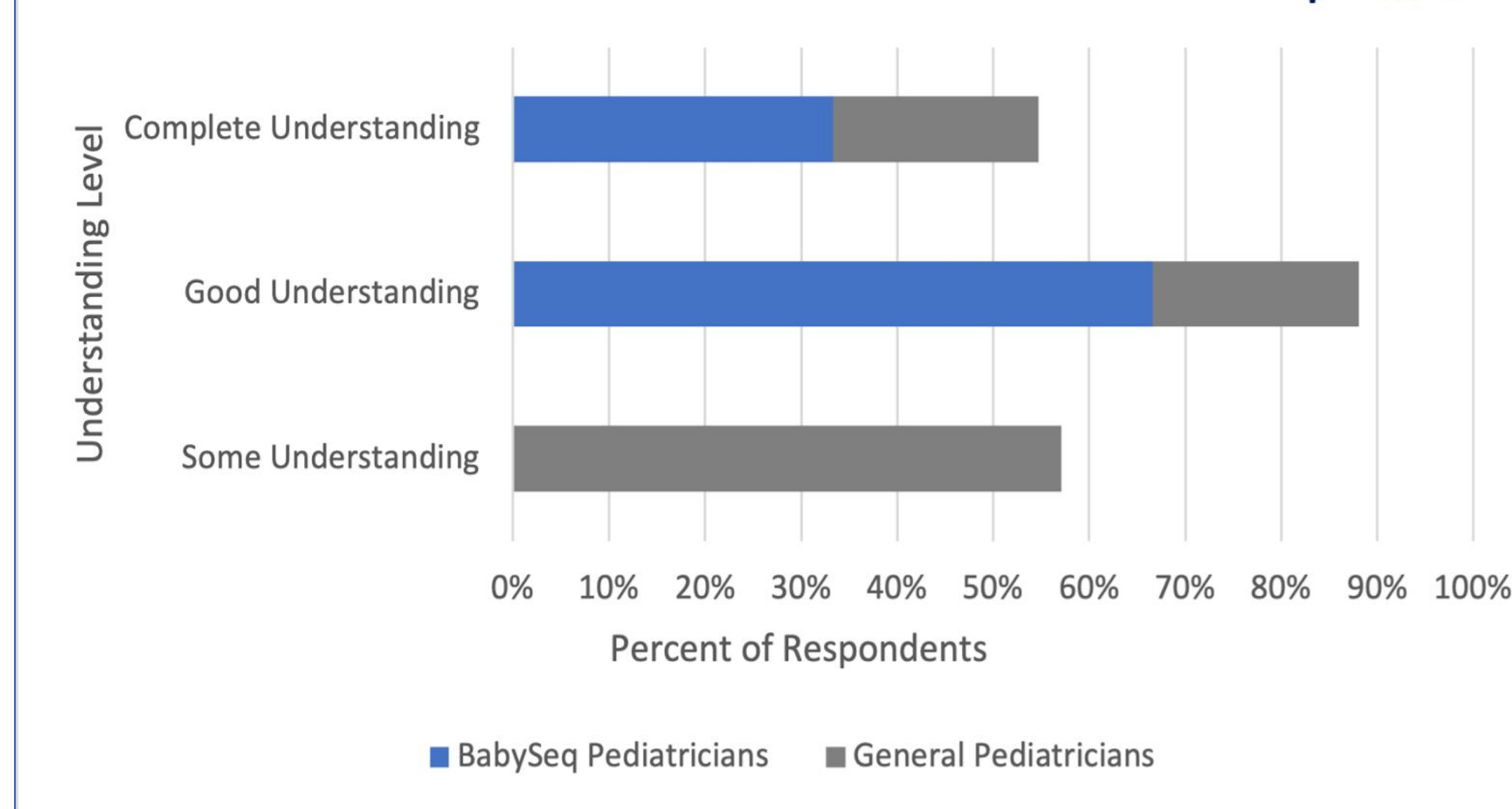
### Pediatrician Understanding towards GS Reports

Section	Average Value
Result Summary	2.83
Childhood-Onset Condition	3.16
Low Penetrance Condition	3.16
Carrier Status	3.05

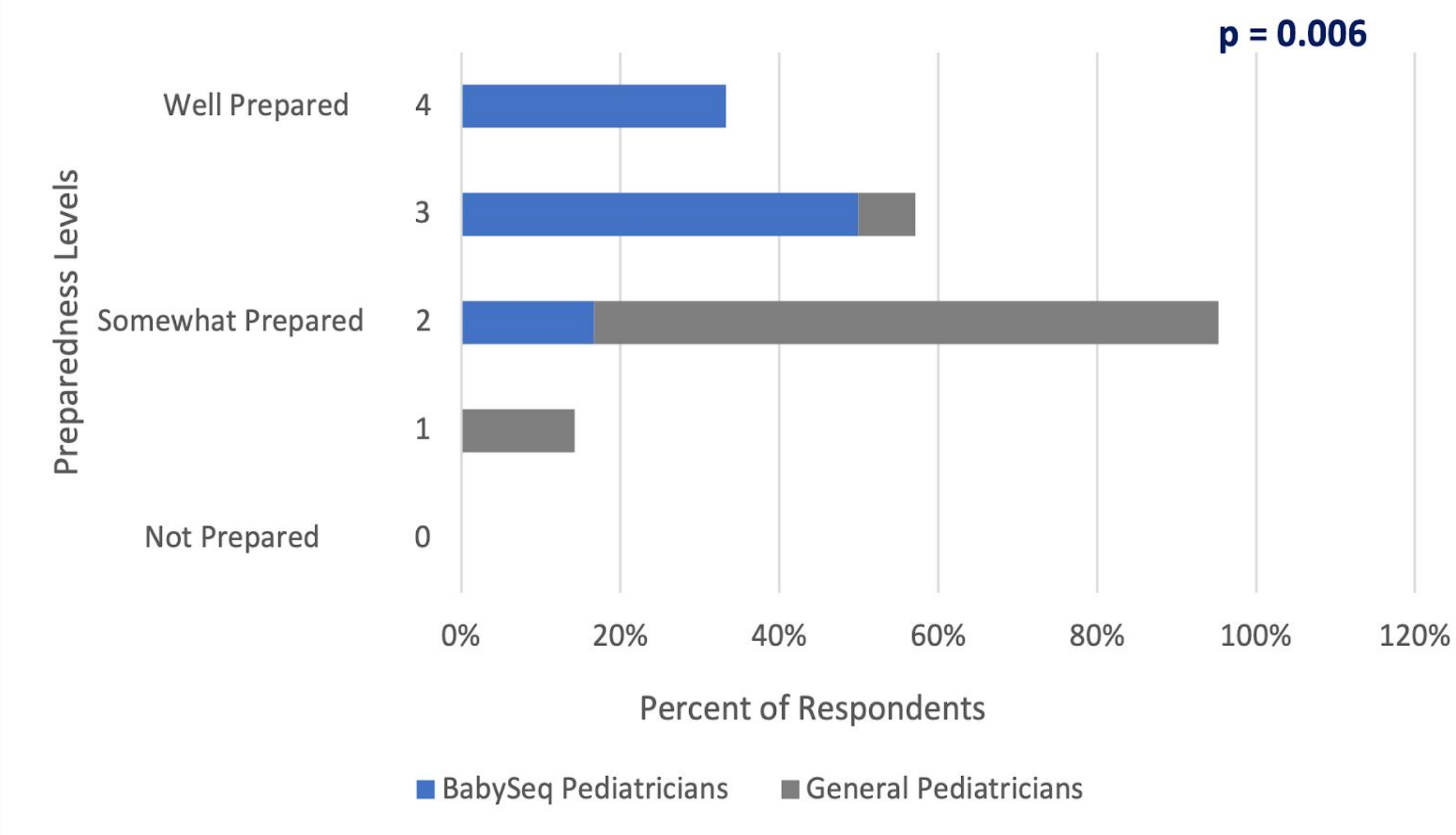
Value	Legend
0	No Understanding
2	Some Understanding
4	Complete Understanding

### BabySeq Pediatricians vs. General Pediatricians and their Reported Understanding - Results Summary



- BabySeq Pediatricians had significantly higher self-reported understanding of the "Results Summary" section of the report compared to General Pediatricians

### Reported Preparedness Levels between BabySeq Pediatricians and General Pediatricians



- BabySeq Pediatricians felt significantly more prepared to interpret GS reports than General Pediatricians
- Only BabySeq Pediatricians felt "Well Prepared"

## Background

- Genome Sequencing (GS) is increasingly being studied for its use as a supplemental Newborn Screening method
- Many primary care physicians lack training in genetics, leading to challenges in interpreting GS results, which may result in misdiagnosis or inappropriate management
- BabySeq addresses these gaps by using GS in newborns and providing genomics training to pediatricians, enhancing their ability to interpret and manage GS screening results effectively

## Methods

- REDCap Survey
- Recruitment through the Babyseq Project and Babyseq recruitment clinics
  - BabySeq Genomics Training: "BabySeq Pediatricians"
  - No BabySeq Training: "General Pediatricians"
- Descriptive and inferential statistical methods used to analyze the data and examine relationships between responses and compared those trained and not trained in genomics through Babyseq

## BabySeq Sample GS Report

- All pediatricians answered questions based on a BabySeq sample GS report for patient "Jane"
- The report had 5 total sections with the following results:
  - Results Summary: Concisely showed all variants detected in each section
  - Childhood-Onset Condition: 2 heterozygous variants for an autosomal recessive condition
  - Adult Onset from the American College of Medical Genetics and Genomics secondary findings list: No variants reported
  - Childhood-Onset with Low Penetrance Condition ("Low Penetrance Condition"): 1 variant for an autosomal dominant condition
  - Carrier Status: 1 variant for an autosomal recessive condition

## Discussion

- Pediatricians were able to comprehend the sample BabySeq GS report, indicating the report's effectiveness in conveying information
  - While some pediatricians had challenges in understanding condition-specific management, most recognized the need for referrals to genetics specialists, regardless of their genetic training background
  - This finding is reassuring due to the increased use of genetic testing results in clinical environments where providers without comprehensive training may be managing findings
- A brief educational program can effectively increase pediatrician's confidence in receiving and managing newborn GS results
- The information in each section of the report is adequate as well as the pediatrician's understanding of each section, supporting the idea that the BabySeq GS report is effective
  - Some individuals commented that their understanding may have increased if recommendation of next steps was more clearly described in the report, which may be helpful to implement

## Implications

- There is a need for expanded genetics education for pediatricians, and targeted training like BabySeq improves understanding and confidence in interpreting reports
- Reports should emphasize the necessity of referrals to genetics specialists, ensuring appropriate patient management
- The challenges pediatricians faced suggest that GS reports should provide more guidance in these areas to prevent mismanagement and enhance patient care