



Diagnostic Timeline and Clinical Efficacy of Telemedicine-Based Pediatric Genetics Care

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Abstract

Background and Objectives: The COVID-19 pandemic necessitated urgent implementation of telemedicine in Pediatric Genetics and Metabolism. While there has been longstanding interest in implementing telemedicine to increase access to Genetics care, questions remain about how remote encounters influence clinical efficacy due to the importance of the dysmorphology physical exam. We studied how telemedicine affected medical care for patients with suspected or confirmed genetic conditions.

Methods: We conducted a retrospective cohort study of 2989 outpatient encounters from April 1, 2020 to October 1, 2020 and 2865 outpatient encounters from the same period in 2019. Visit types, diagnoses, patient demographics and laboratory data were acquired from the electronic health record, and patient satisfaction was assessed using Press Ganey scores.

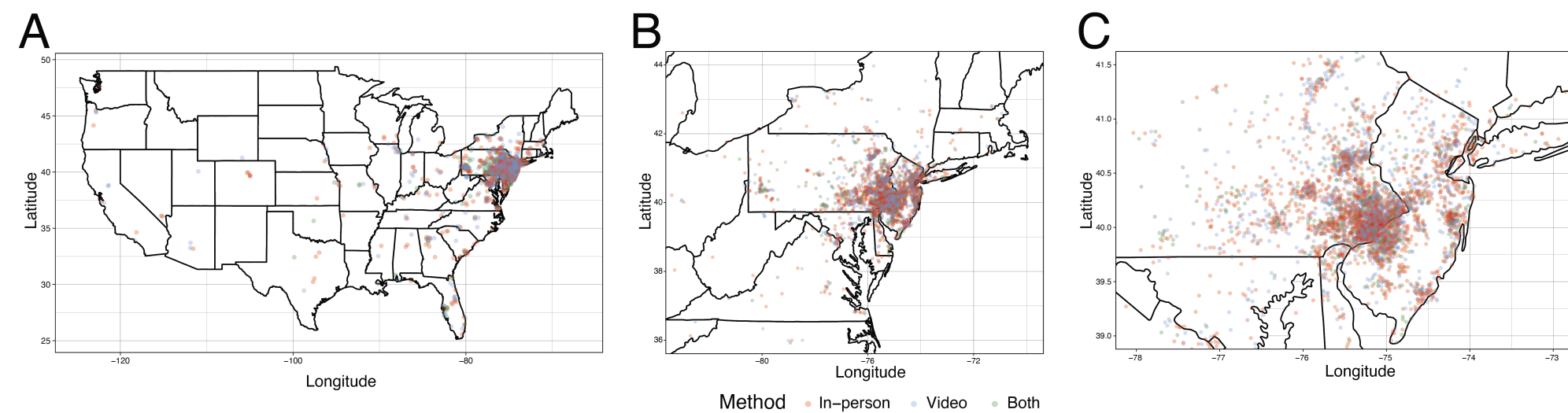
Results: Comparing patients receiving virtual and in-person evaluations, we found significant differences in age, race and ethnicity, preferred language, and income. Rates of overall patient satisfaction were similar. Among patients undergoing evaluation for suspected genetic disease, providers intended to perform more genetic testing for patients evaluated virtually. However, patients seen in person were more likely to have a DNA sample drawn thereby resulting in similar test completion rates. Ultimately, there was no significant difference in molecular diagnosis rate for in-person versus virtual evaluation.

Conclusions: We found virtual evaluation to be non-inferior to in-person evaluation from a clinical efficacy perspective, but improved methodologies for remote sample collection may be required. Our results are undoubtedly influenced by particularities of our clinical workflow but are nonetheless relevant to other specialties with perceived importance of physical examination.

Patient Demographics

	In-person Only (n = 2642)	Video Only (n = 1685)	Total (n = 4883)	p-value
Age (years)				< 0.001
Mean (SD)	8.307 (9.280)	8.795 (11.370)	8.789 (10.505)	
Range	0.014 - 72.5	0.004 - 76.9	0.004 - 76.9	
Sex				0.973
Female	1216 (46.0%)	782 (46.4%)	2258 (46.2%)	
Male	1425 (53.9%)	902 (53.5%)	2623 (53.7%)	
Race / Ethnicity				< 0.001
Hispanic or Latino	281 (10.6%)	200 (12.0%)	528 (10.9%)	
Non-Hispanic Black	299 (11.3%)	146 (8.8%)	479 (9.8%)	
Non-Hispanic White	1574 (59.6%)	1034 (62.0%)	2994 (61.6%)	
Other	485 (18.4%)	288 (17.3%)	862 (17.7%)	
Preferred Language				< 0.001
Arabic	25 (1.0%)	2 (0.1%)	29 (0.6%)	
English	2429 (92.5%)	1573 (95.0%)	4529 (93.7%)	
Spanish	105 (4.0%)	60 (3.6%)	183 (3.8%)	
Other	66 (2.5%)	21 (1.3%)	94 (1.9%)	
Median Home ZIP Code Income (USD)				0.007
Mean (SD)	83336 (38957)	87160 (40546)	84769 (39297)	
Range	13235 - 250001	16607 - 250001	12500 - 250001	
Distance (km)				0.188
Mean (SD)	118 (312)	133 (351)	126 (323)	
Range	0.879 - 4047	0.952 - 4027	0.879 - 4047	
Payor Type				< 0.001
Commercial	1628 (61.6%)	1095 (65.0%)	3085 (63.2%)	
Medical Assistance	642 (24.3%)	468 (27.8%)	1205 (24.7%)	
Other	372 (14.1%)	122 (7.2%)	593 (12.1%)	

Patients Evaluated by Our Practice Come from Surprising Distances, But There Was No Significant Difference Between Visit Methods



Geographic distribution of patients across **A)** the United States, **B)** the northeast and **C)** the tri-state area. Red circles denote patients seen through in-person encounters, blue circles denote patients seen through telehealth encounters and green circles denote patients seen through both visit types. Random noise was introduced for patient privacy.

Family Satisfaction

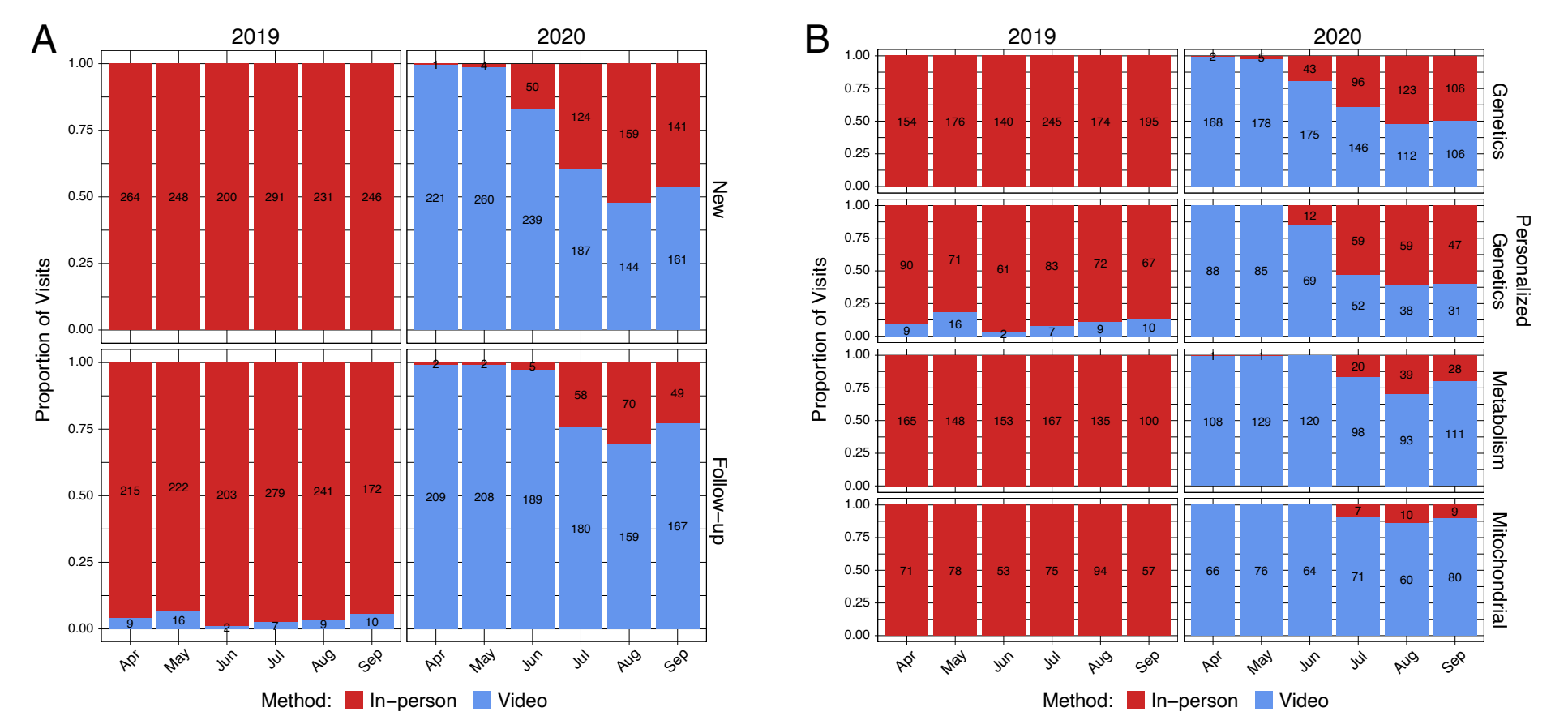
	Very Poor, Poor, or Fair		Good		Very Good	
	2019	2020	2019	2020	2019	2020
Overall Assessment (%)	1.6%	2.1%	9.2%	9.5%	89.2%	88.4%
n	10	4	56	18	543	168
Access (%)	10.9%	3.2%	28.3%	22.5%	60.8%	74.3%
n	47	6	122	43	262	142
Moving Through the Visit (%)	18%	15.4%	24.7%	19.3%	57.3%	65.3%
n	82	23	113	29	262	98

Note: Survey results were not stratified by visit method. Not all questions were asked of all respondents.

References

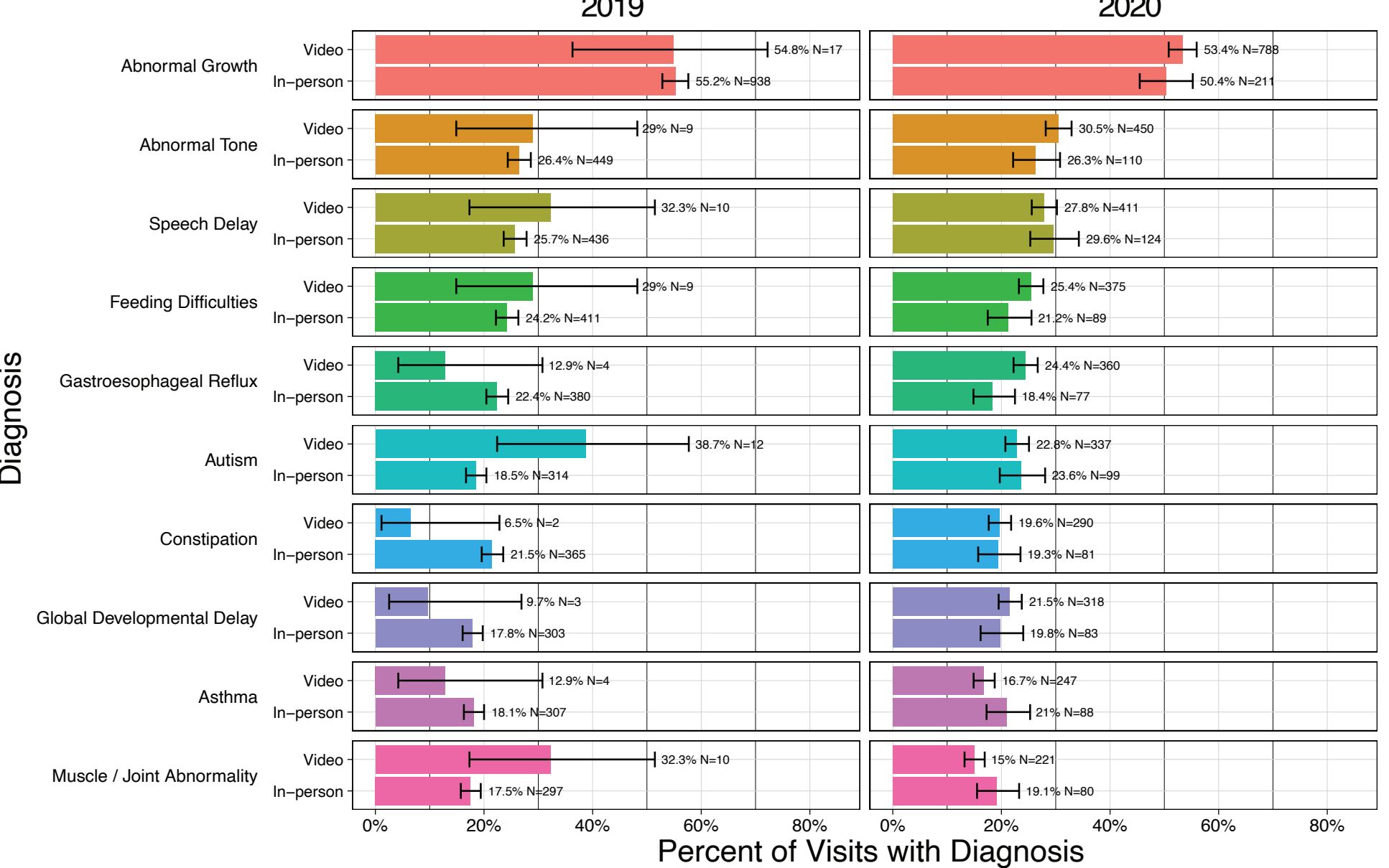
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Clinical Genetics Returned to In-person Evaluation Sooner, Potentially Indicating Perceived Importance of the Physical Exam



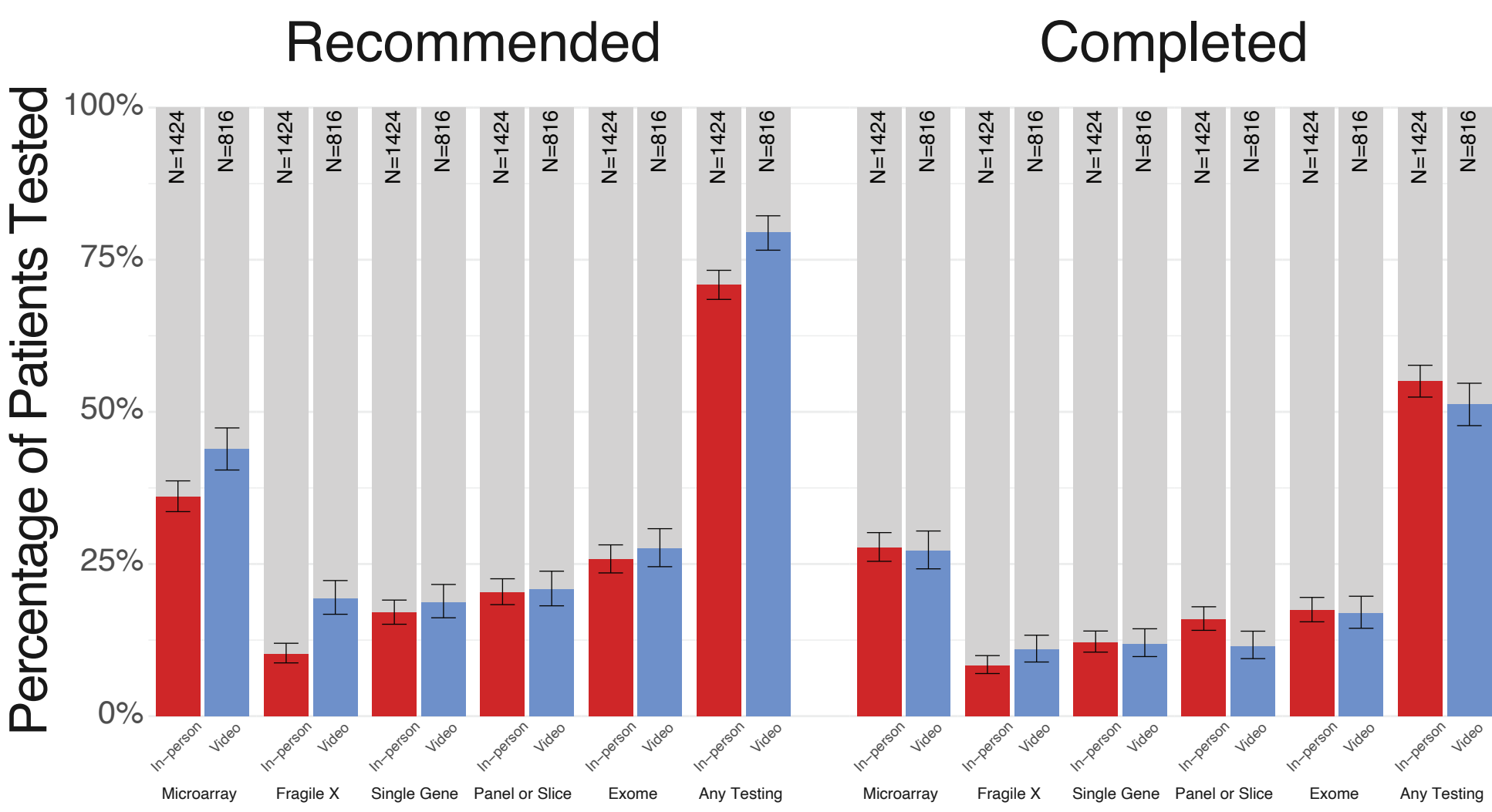
A) Distribution of in-person versus video visits for new and follow-up appointments across the Division of Human Genetics. **B)** Distribution of in-person versus video visits for each section within the Division of Human Genetics.

Visit Diagnoses Were Not Materially Different Between Visit Methodologies



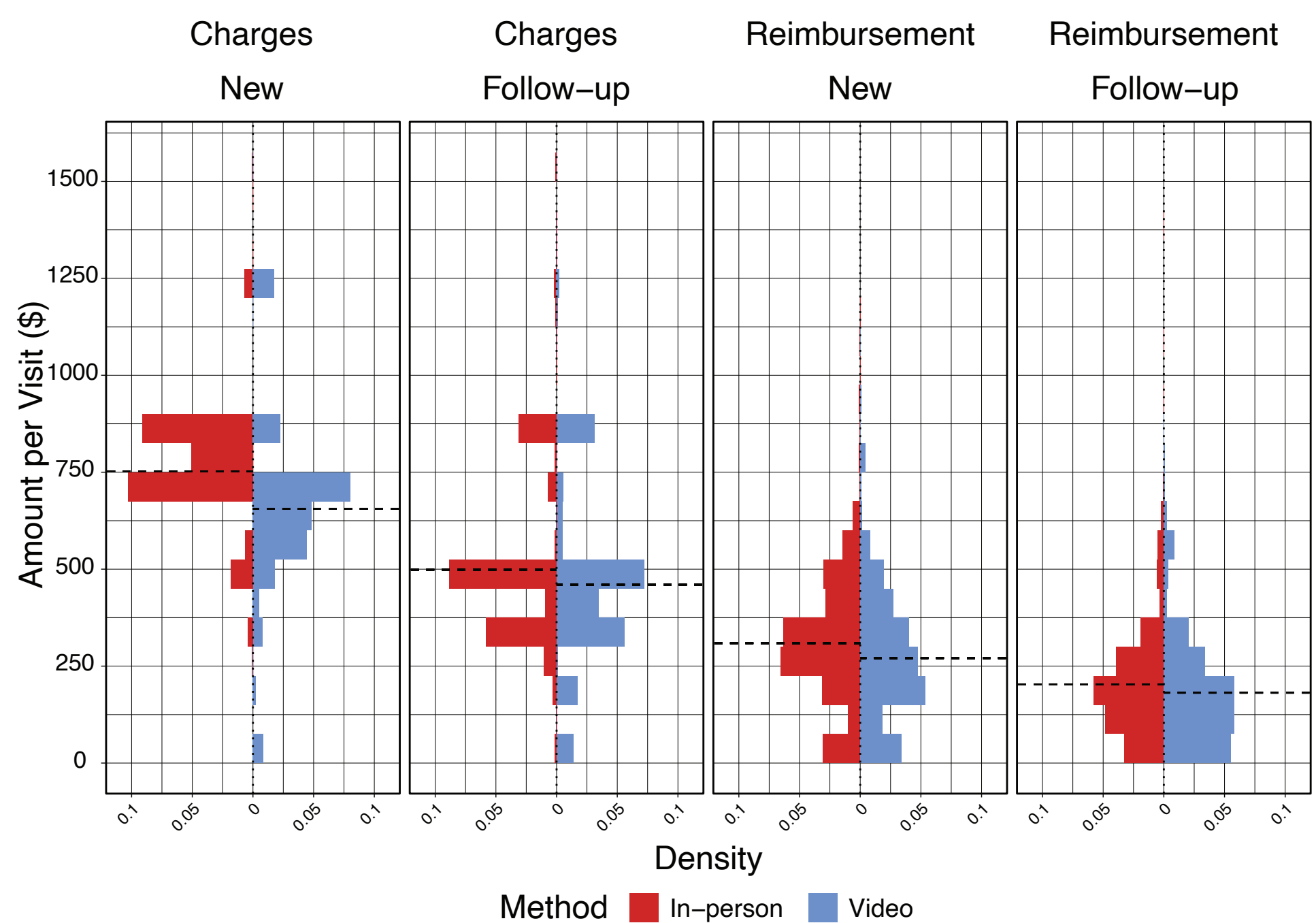
Distribution of the 10 most frequent diagnoses aggregated by ICD-10 category. Error bars indicate the 95% confidence interval of the proportion.

Clinicians Intended to Perform More Testing When Initial Evaluation Was by Video, But Actually Completed Testing Was Similar



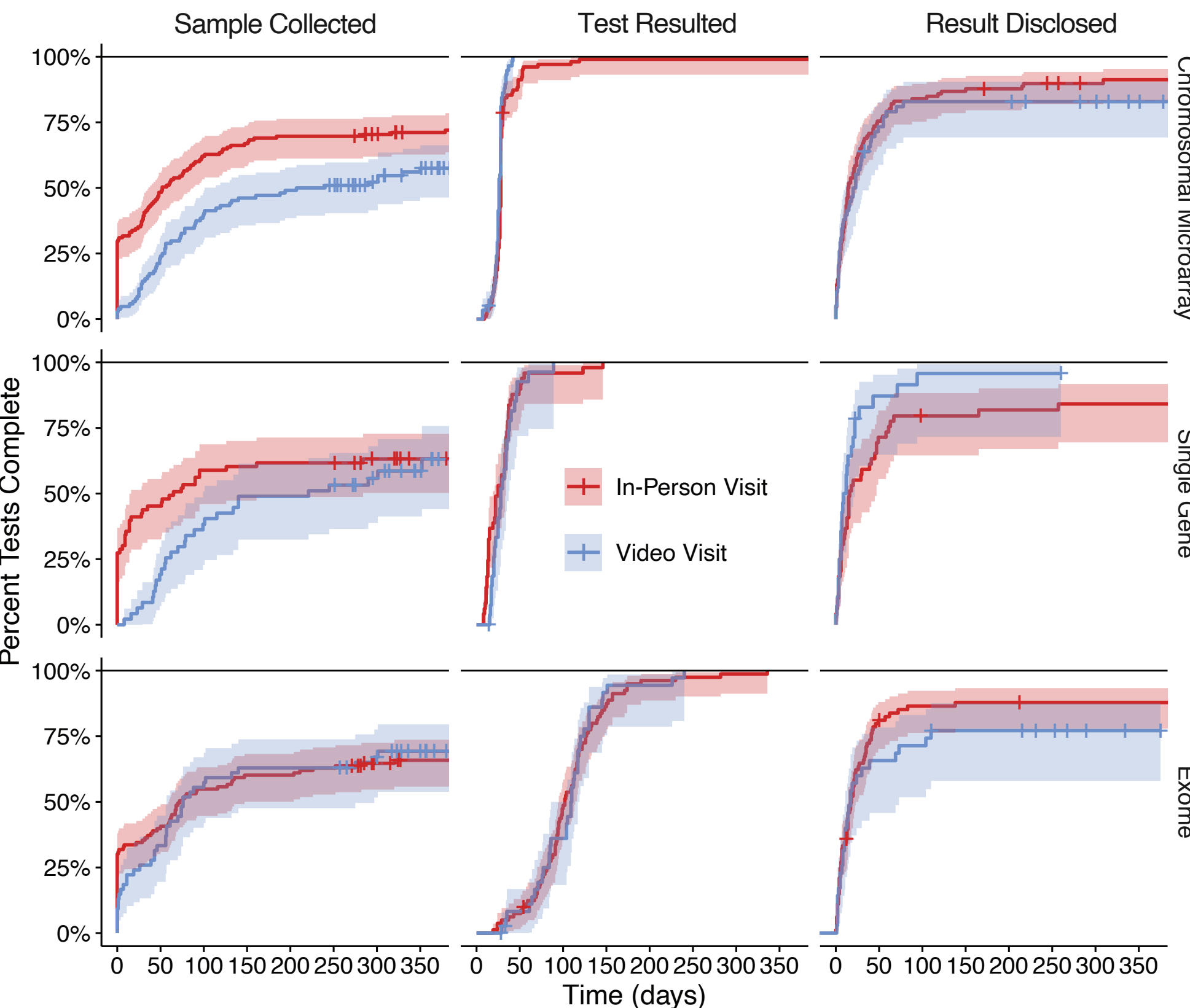
Percentage of patients recommended to undergo a given diagnostic test and percentage completed. Completion was defined as documentation of results return in the electronic health record. Error bars indicate the 95% confidence interval of the proportion

Although Charges Generated for New Video Visits Were Lower, Ultimate Reimbursement Was Less Affected



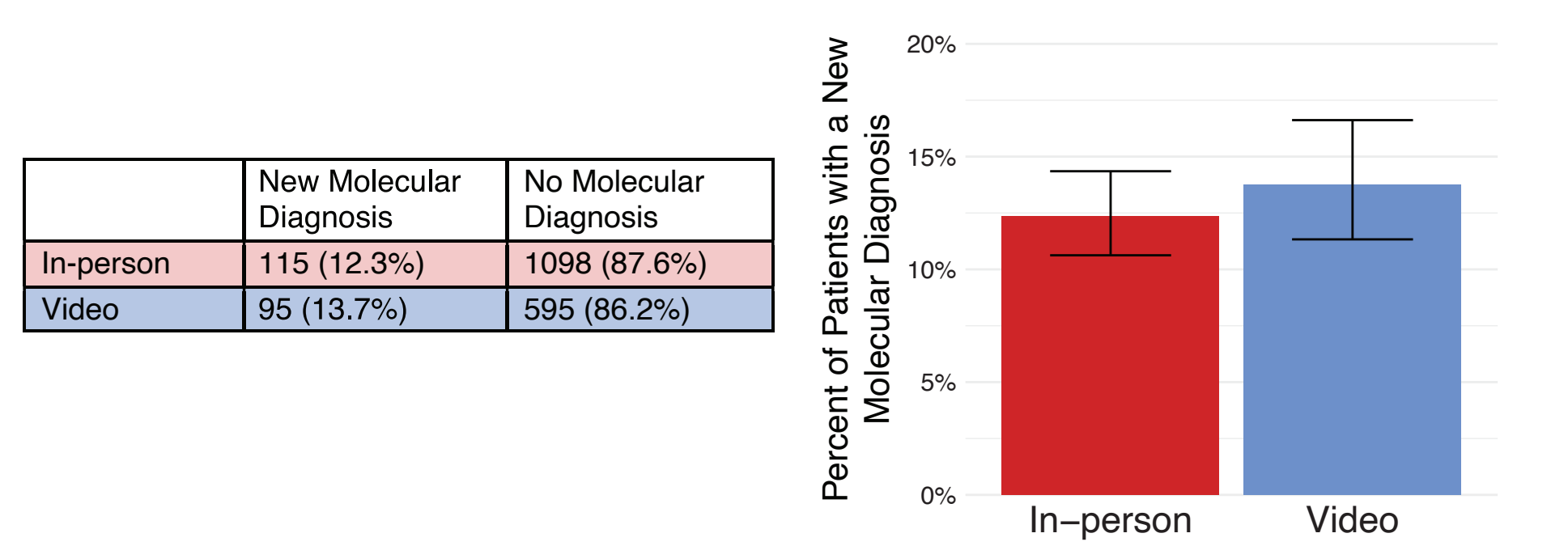
Charges were on average \$98 less for video new patient visits in comparison to in-person evaluation. Follow-up video visits generated \$39 less in charges on average. Meanwhile, reimbursement was \$38 and \$22 less, respectively. Together, this represented a total of \$72,186 of potentially lost revenue over the 6 month study period in 2020.

A Lower Percentage of Recommended Testing Was Completed for Patients Evaluated by Video, Mostly Mediated by Decreased Sample Collection on the Day of the Visit



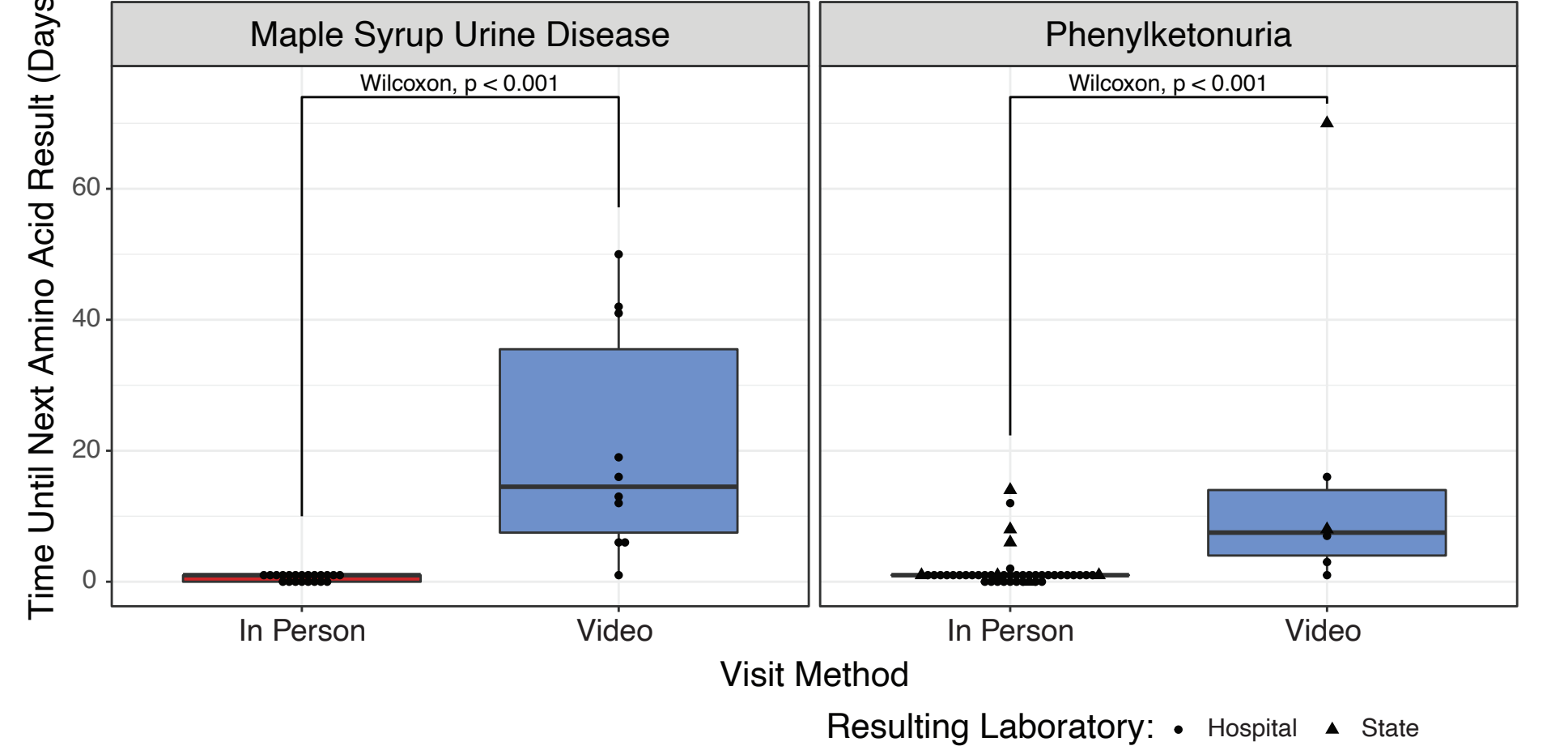
Diagnostic timeline of genetic testing for three test classes of interest. “Sample collected” represents the time elapsed between the day the test was recommended and the day the sample was documented as received by our hospital’s lab or the outside reference lab. “Test result” indicates the time elapsed between sample collection and the date of the report. “Result disclosed” indicates the time elapsed between the date of the report and the date the results disclosure was documented in the EHR.

The Rate of New Molecular Diagnosis Was Non-inferior for Patients Evaluated Initially by Video



Among new patients without a pre-existing diagnosis evaluated by our Clinical Genetics sections, we found initial video evaluation to be non-inferior to in-person evaluation from a new molecular diagnosis perspective (13.8% vs 12.4% respectively, p = 0.40, Fisher’s exact test).

Video Evaluation Was Associated with Delayed Metabolism Monitoring Labs



Time between metabolism visit and the next routine monitoring amino acids result was significantly delayed for both Maple Syrup Urine Disease (median 15 day longer delay) and Phenylketonuria (PKU, median 6.5 day longer delay) when evaluation was by video. Circles indicate results from our hospital metabolic lab while triangles indicate results from the state newborn screening labs (only available for PKU). The lesser delay seen among PKU monitoring labs may be due to family familiarity with remote sample collection.

Conclusions

- Patients evaluated exclusively by telemedicine were more likely to identify as white, speak English as their preferred language, live in more affluent neighborhoods, and have commercial insurance compared to patients evaluated exclusively in-person.
- Clinical Genetics sections returned to in-person evaluation sooner than other sections potentially due to perceived importance of the dysmorphology physical exam.
- Clinicians intended to perform more genetic testing for patients evaluated by video, but due to problems with sample collection, rates of actually completed testing were similar between modalities.
- The clinical efficacy (actually arriving at a new molecular diagnosis) of initial video evaluation was non-inferior to in-person evaluation.
- Charges and reimbursement for video visits were slightly lower compared to in-person evaluations.