Barriers to genomic medicine implementation as identified by focus and related programs; “XX” indicates critical barrier.

Barriers Identified	Focus Programs								Related Programs													
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GAPH	GTEx	GS-IT	IOM	ISCC	LSAC	MVP	PAGE	PCORNet	PhenX	PGRN
DATA/INFORMATION NEEDS																					
Evidence base for implement’n incl long-term outcomes			X	X			X	X						X							
Common data elements	X					X						X		X					X		
Development, validation of phenotypes			X	X															X		
Specific drug response phenotypes to add to trials																					X
Publicly available genotype/phenotype info						X			X			X									
Framework for classifying/curating actionable variants			X			X															
Unclear penetrance of actionable genes			X	X		X															
Frequency, impact of variants in ancestrally diverse pop’ns			X	X		X												X			
Loss of interoperability btwn sequence data repositories													X								
RAPIDLY EVOLVING NATURE OF GENOMIC INFO																					
Evolving molecular testing panels					X																
Changes in evidence and subsequent treatment		X	X		XX																
Division between discovery and implementation																X					
CLINICAL IMPLEMENTATION ISSUES																					
High cost of sequencing, data processing								X									X				
Targeted testing vs genome-scale sequencing			X																		
Limited use of standardized EMR terms, ontologies				X		X															
Concise, comprehensive, interoperable lab reports			X							X											
Integration of genomic data in learning healthcare system			X	X																	
Turnaround in clinically emergent settings			X																		
Use cases for genomic CDS development										X											
Limited usefulness and interoperability of CDS				X	X		X	X													
Rapidly evolving EMRs					XX																
Limited transportability of clinical workflows, protocols			X		XX																
Differing education needs across professional levels					X																
Returning incidental findings				X								X									
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM	G2MC	CMG	CPIC	GAPH	GTEX	GS-IT	IOM	ISCC	LSAC	MVP	PAGE	PCORNe	PhenX	PGRN

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REGULATORY NEEDS																					
Central IRB	X																				
Sharing identifiable data across collaborating sites	X					X															
Privacy threats (FISMA compliance for UDN)	X			X																	
Regulations impeding return of results	X	XX	X																		
Need for cloud computing																X		X			
Reimbursement policies and regulations	XX		X																		
OTHER																					
Lack of centralized iPS cell repository												X									
Installing and using sequence analysis software													X								
Different tools for variant calling give different results													X								
Finding and interpreting CNVs and SVs													X								
Difficulty focusing on specific genes in massive BAMs													X								
Need secure systems enabling access by many users						X										X					
Harnessing social media and crowdsourcing methods						X													X		
Increasing visibility and use of tools; tracking uptake						X													X		
Need for bedside back to bench research				X			X														
Publication standards for rapid research results						X			X												
Worldwide ascertainment of Mendelian families									X												
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GAPH	GTEX	GS-IT	IOM	ISCC	LSAC	MVP	PAGE	PCORNet	PhenX	PGRN