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HARVARD MEDICAL SCHOOL

DECEMBER 1, 2023

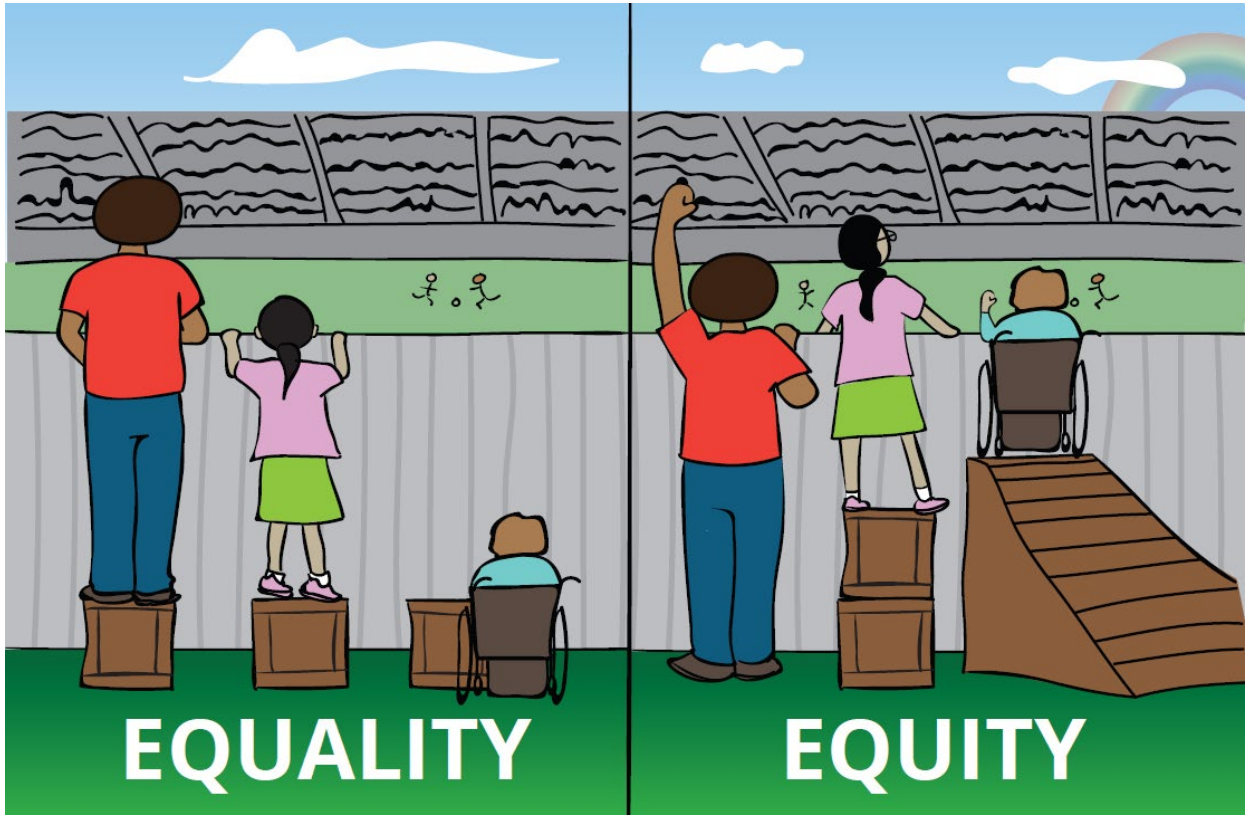
NEW ENGLAND CONSORTIUM OF METABOLIC PROGRAMS:

EQUITABLE ACCESS TO CARE

OUTLINE

1. Health Equity
2. Delayed Genetic Diagnoses in Pediatric Patients

HEALTH EQUITY



(EquityTool, 2023)

HEALTH EQUITY

“Equity is the absence of unfair, avoidable or remediable differences among groups of people, whether those groups are defined socially, economically, demographically, or geographically or by other dimensions of inequality (e.g., sex, gender, ethnicity, disability, or sexual orientation). Health is a fundamental human right. Health equity is achieved when everyone can attain their full potential for health and well-being.”

- World Health Organization

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

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Background Information

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  - 20,466 pediatric referrals over one year in a 52-center network
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    - >7 day interval delay

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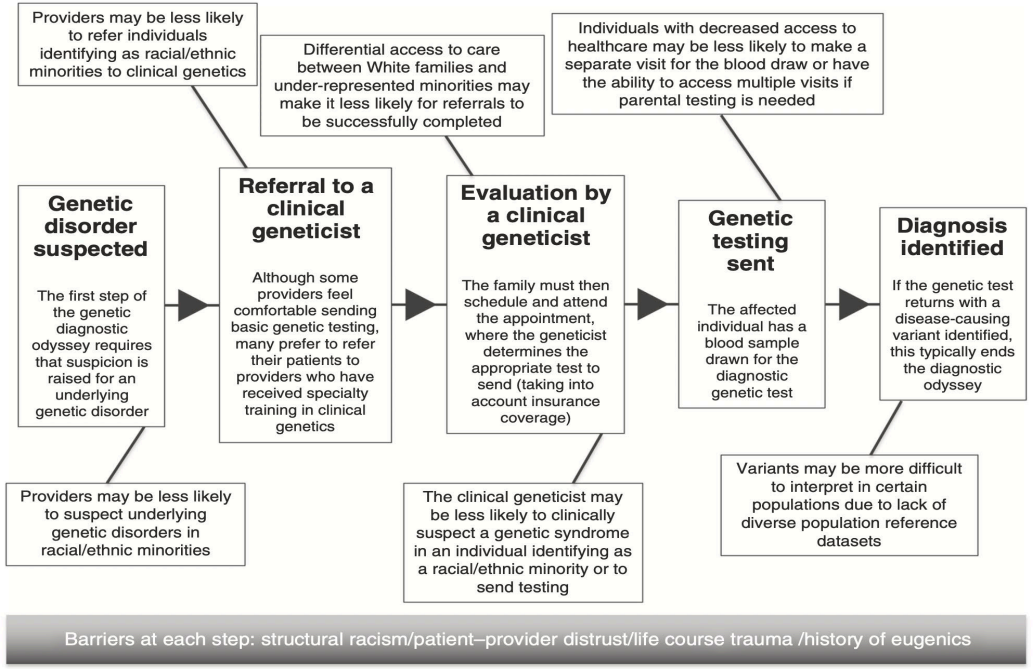

Fig. 1 Barriers along the path to a genetic diagnosis. Illustrating the process of identifying a genetic diagnosis for individuals with rare disease and the possible interference of social determinants of health.

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Objective

- Identify cases of delayed genetic and metabolic diagnoses for pediatric patients from minority populations and assess the contributors to the delay.

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Hypothesis

- There will be multiple contributors to delayed diagnoses.

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Experimental Design

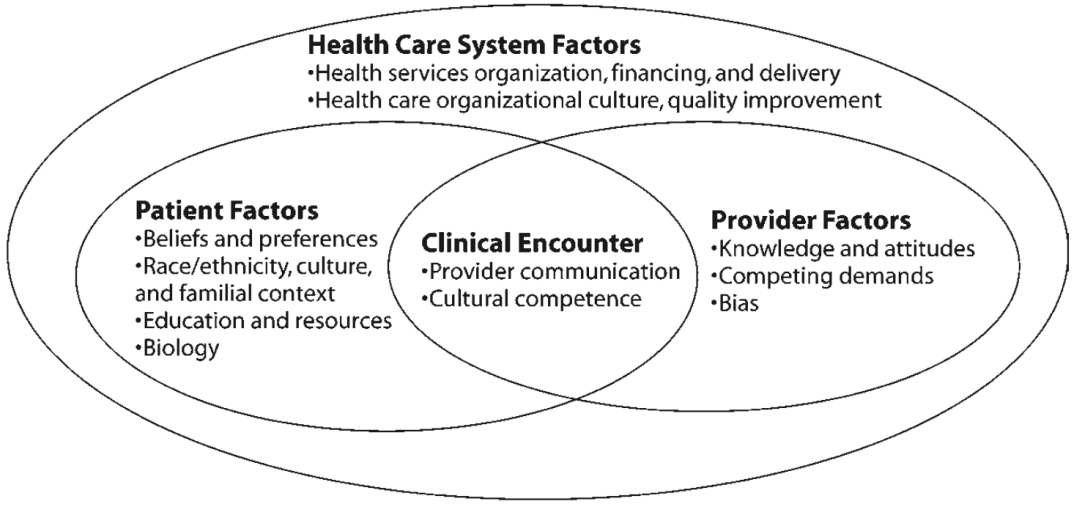

FIGURE 2—Understanding the origins of health and health care disparities from a health services research perspective: key potential determinants of health disparities within the health care system, including individual, provider, and health care system factors.

(Kilbourne et al., 2006)

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Case 1: Health System Factors

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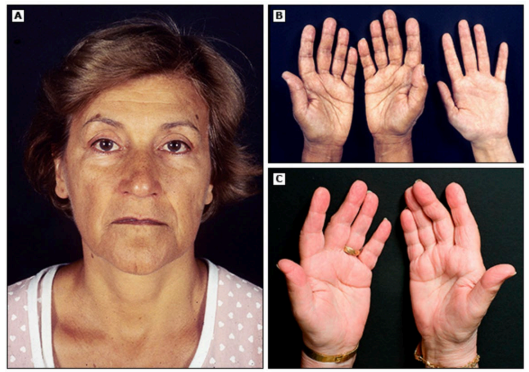
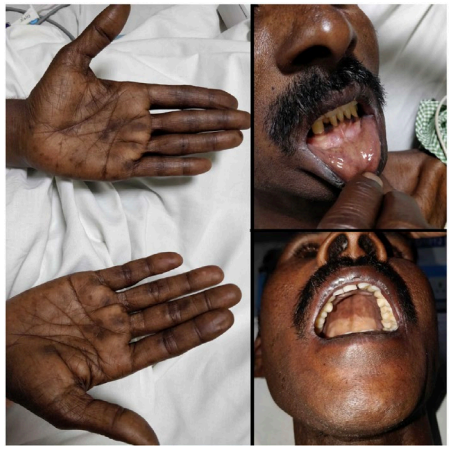
- Contributors to delay:
  - Limited reference materials



(Nieman, 2023)



(Arul et al., 2019)

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- Contributors to delay:
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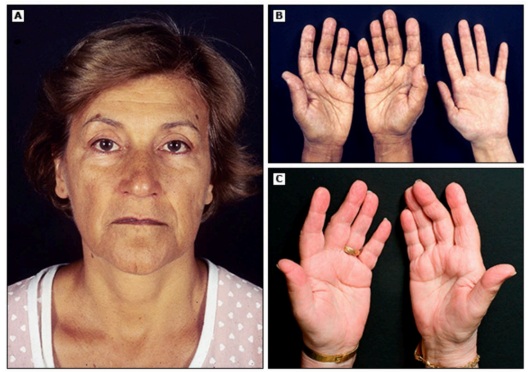
  - Sequencing with decreased diagnostic rate



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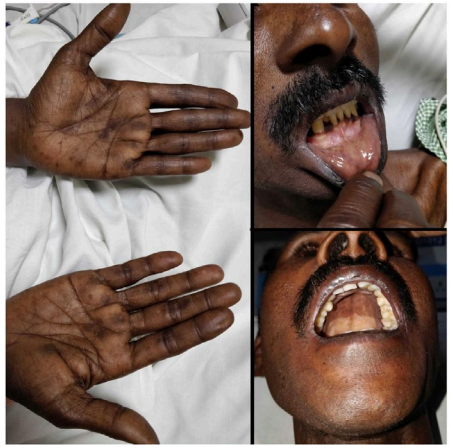

(Arul et al., 2019)

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Case 2: Provider Factors

- Patient:
  - 2 year old African American girl with microcephaly, referred to Neurology
  - Exam notable for developmental delay with normal brain MRI, referred to Genetics
  - Dysmorphic features and stereotyped behavior appreciated on exam

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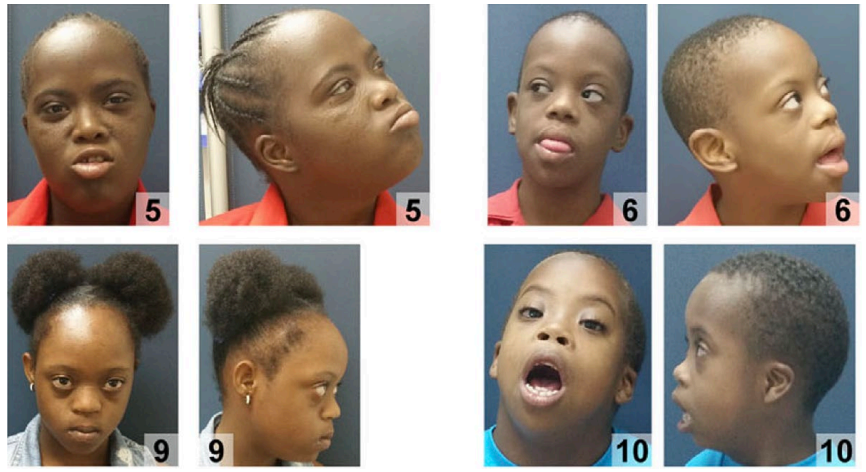

(Kruszka et al., 2016)

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- Contributors to delay:
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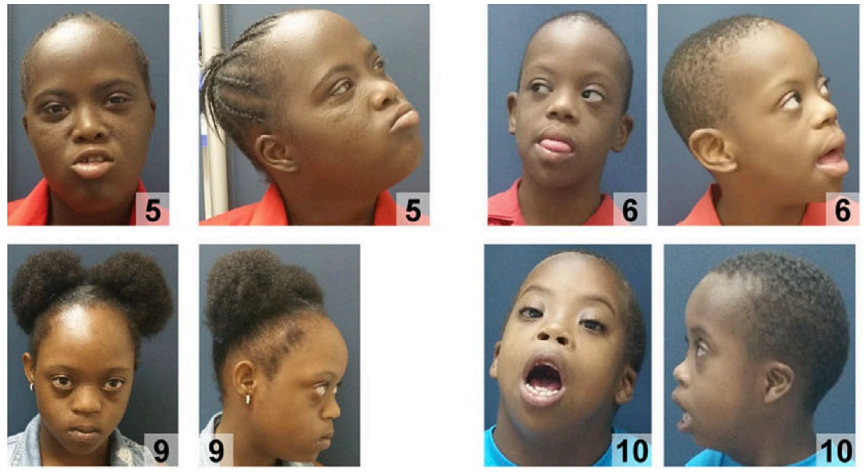
  - Low suspicion for genetic disease



(Kruszka et al., 2016)

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Case 3: Patient Factors

- Patient:
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  - Non-diagnostic genetic and metabolic evaluation, lost to follow-up at 7 years old
  - Presented with pneumonia and failure to thrive at 13 years old

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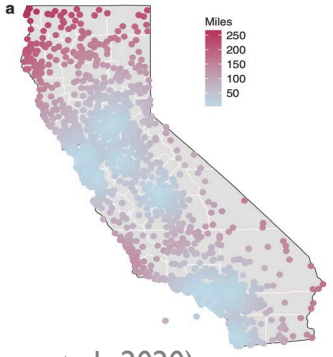
- Diagnosis: Glycogen Storage Disease, Type IXa

- Contributors to delay:
  - Lost to follow-up
  - Poor geographic access
  - Lack of health insurance

Distance to genetics health-care services in California.



(Penon-Portmann et al., 2020)

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

Cases of delayed genetic diagnosis due to health system, provider, and patient factors.

Factor	Diagnosis	Age at Diagnosis	Average Age of Diagnosis for this Condition	Perceived Reason for Delay
Case 1: Health System	X-linked Adrenoleukodystrophy	7 years old	6.8 years old	• Physical exam finding dismissed in African American patient
Case 2: Provider	Down Syndrome	2 years old	95% diagnosed prenatally	• Focus on one anomaly • Dysmorphic features overlooked in African American patient • Geographic isolation
Case 3: Patient	Glycogen Storage Disorder Type IXa	13 years old	2.6 years old	• Loss to follow-up • Incomplete scope of genetic testing at time of initial presentation

(Shapiro et al., 2000; Hobson-Rohrer et al., 2013; Liang et al., 2020)

DELAYED GENETIC DIAGNOSES IN PEDIATRIC PATIENTS

ORIGINAL ARTICLE

AMERICAN JOURNAL OF medical genetics PART A WILEY

Delayed diagnosis and racial bias in children with genetic conditions

Jacklyn Omorodion[1,2,3] | Leah Dowsett[4,5] | Robin D. Clark[6] | Jamie Fraser[7] | Aya Abu-El-Haija[8,9,10] | Alanna Strong[11] | Monica H. Wojcik[8,10,12] | Allison S. Bryant[13] | Nina B. Gold[10,14]

DELAYED DIAGNOSIS AND RACIAL BIAS IN PEDIATRIC PATIENTS WITH GENETIC CONDITIONS

Table 1. Suggestions to address the health system, provider, and patient factors that contribute to delays in genetic diagnoses for pediatric patients.

Health System Factors

- Include examples of physical exam findings in individuals of different ethnicities and skin tones in reference texts, manuals, and databases.
- Incorporate virtual clinics into the clinic model and optimize telemedicine capabilities for patients that may be geographically isolated from specialty clinics, such as genetics clinics.
- Provide financial support for medically indicated genetic testing for all patients.
- Develop pathways for enrollment in clinical trials and federally sponsored testing and research programs that are accessible to patients from racial minority, non-English speaking, low literacy, and/or low socioeconomic groups.
- Hire a work force that is representative of the patient population served as representation matters and impacts the patient-provider relationship.

Provider Factors

- Consider genetic conditions when developing differential diagnoses, especially in children with dysmorphic features, multiple congenital anomalies, and/or intellectual disability of unclear etiology.
- Acknowledge that implicit biases may be present and aim to mitigate the impact this can have on engagement with families and discussions about diagnoses, management, and research opportunities.
- Participate in cultural humility training that is now available, if not required, at many institutions.

Patient Factors

- Educate patients about the role of medical genetics in their evaluation and the reason for referral, explaining it is more than just research or an academic pursuit.
- Combat distrust by acknowledging the troubling history of genetics in medicine and addressing patient worries about this when expressed.
- Elicit patient concerns about genetics referrals and discuss any reservations they may have.
- Invite patients to bring family members and/or trusted colleagues to these, at times, difficult conversations with implications for others.

(Omorodion et al., 2022)

THANK YOU

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QUESTIONS?