



The Role of Education: Addressing Challenges and Opportunities to Clinical Translation

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Drivers of Genomic Medicine

Clinical utility

- Diagnostic (including closure to diagnostic odysseys)
- Management (syndromic versus non-syndromic, PGx)
- Assessing risk to family members

Market forces

- Increasing availability of FDA-approved tests and LDT tests
- Competition: advertise the “practice of precision medicine”

Patient expectations

- Research shows that patients expect providers to understand genomics and that this is a source of anxiety for providers.

Top-down pressure

- Some integrated health systems are beginning to mandate/regulate clinical pathways involving genomics as a means of improving care and saving money.

Barriers to Genomic Medicine

Clinical utility/workflows

- Paucity of evidence
- Uncertainty
- EMR/CDS

Clinical guidelines

Payer issues- e.g., Cigna, Tricare

Skill and knowledge gaps

Access

Access to Genetics Expertise

Best: academic medical centers/tertiary care

Worst: rural areas; underserved urban centers;
community health settings

Specialist Providers

MD clinical geneticists: 1,500 (ABMG); 14 states have 5 or fewer

CGCs: 3,500 (ABGC); an increasing number work in industry

Neurologists: 16,300, with a looming shortage forecasted (AAN)

PCPs: 430,000 (KFF)

Reality: Non-genetics specialists will increasingly practice genomic medicine. Need to start preparing them now.

Genetic Testing Guidelines in Neurology

Screening and Diagnosis of Autism

Clinical practice recommendations

1. Genetic testing in children with autism, specifically high resolution chromosome studies (karyotype) and DNA analysis for FraX, should be performed in the presence of mental retardation (or if mental retardation cannot be excluded), if there is a family history of FraX or undiagnosed mental retardation, or if dysmorphic features are present (Standard). However, there is little likelihood of positive karyotype or FraX testing in the presence of high-functioning autism.

Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Child Neurology Society, Filipek, PA, *et al.*, 2000, reaffirmed in 2014

Global developmental delay (2011)

Distal symmetric polyneuropathy (2009)

Epilepsy?

Educating Providers about the Genetic Basis of Disease: Focus on Common Skills

Assessing genetic risk

Identifying red flags, patterns, and stratifying risk

Communicating benefits and limitations of genetic testing

Interpreting and communicating test results

Using guidelines and managing in the context of genetic risk

Opportunities and Challenges for Education

UG medical education

- basic science vs. clinical application
- time gap to impact
- parochial curriculum; role of role of USMLE

GME: preceptors, trainees, and the system

- NHGRI: G2C2, ISCC

CME - PI-CME: time consuming, difficult, unclear efficacy

- MOC: general backlash
- common, familiar, easiest to implement

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CME

- PI-CME: time consuming, difficult, unclear efficacy
- MOC: general backlash
- potential for rapid impact; large audience (e.g., PCPs)
- ability to capitalize on IPE

Opportunities and Challenges for Education

Emphasize skills over knowledge

- The importance of family history, risk assessment, patient communication, and when to refer.

Funding

Developer: commercial vs. professional society