The NHGRI Short Course in Genomics: Energizing Genetics and Genomics Education

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91 educators from middle schools, high schools, community colleges, and tribal colleges

28 states and 2 territories represented
A Randomized Controlled Trial of an Online Health Tool About Down Syndrome

**Question:** Does a novel online health tool, Down Syndrome Clinic to You (DSC2U), improve adherence to national Down syndrome guidelines, and are primary care providers (PCPs) and caregivers satisfied with it?

In a national randomized clinical trial of 230 caregivers with children or dependents without access to a DS specialist:

- **1.6x MORE EVALUATIONS**
  - The intervention group had a 1.6-fold increase in the number of evaluations recommended or completed compared to controls.

- **Nearly all PCPs agreed with the recommendations in the plans, and both caregivers and PCPs reported high levels of satisfaction.**

**Bottom Line**

DSC2U improved adherence to national Down syndrome healthcare guidelines and was highly valued by caregivers and PCPs.

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**Importance:** Down Syndrome (DS) clinics are geographically inaccessible for many; greater than 33% of patients with DS would need to drive more than 2 hours to reach the nearest clinic. As such, the majority of people with DS are likely not receiving adequate evidence-based screening and preventive care, resulting in delayed, missed, or undertreated comorbidities.

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Clinical impact of genomic testing in patients with suspected monogenic kidney disease

Methods & Cohort
- 4 tertiary hospitals in Melbourne, Australia
- 204 patients with clinically suspected GKD
- Assessment in multidisciplinary Renal Genetics clinic and exome sequencing

Results
- 80/204 (39%) – diagnosis
- 47/80 (59%) – changed management
- 40/80 (50%) – informed cascade testing

- 35/80 (44%) changed surveillance
- 16/80 (20%) changed treatment
- 10/80 (13%) avoided biopsy

Younger age at presentation and positive family history was predictive of diagnosis through exome sequencing

Conclusions In a pediatric and adult cohort with suspected monogenic kidney disease, ES had high diagnostic and clinical utility. Our findings, including predictors of positive diagnosis, can be used to guide clinical practice and health service design.

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