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Detecting Unaffected Individuals with Lynch Syndrome (DUAL)

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DISCLOSURES

Full-time employee at UT Southwestern Medical Center

No financial conflicts of interest
Scope of the Problem

- Annual Colon Cancer Treatment Costs:
  - $14 billion nationally
  - $3.7 billion in Texas

- Lynch syndrome:
  - Prevalence: 1 in 300
  - ~5% of colorectal cancers
  - Only ~3% identified

Risser DR et al. Tex Med 2010
National Cancer Institute https://doi.org/10.1093/jnci/djt021
Lynch Syndrome (LS) Cancer Risks

*Other Lynch syndrome cancers: urinary tract, bile duct, small bowel, brain, pancreas, sebaceous neoplasms

**Breast and prostate cancers

Barriers to Identifying LS

- Strict testing criteria (Amsterdam & Bethesda)
- Testing focused on affected individuals
- LS is often unrecognized by physicians
- Inaccurate reporting of family history by the patient
Disparities in Access to Genetic Health Services

Minority and geographically isolated groups experience significant disparities in access to cancer genetics services.

**RURAL GEOGRAPHIC ISOLATION**

**EDUCATION**

**SOCIOECONOMIC STATUS**

**CULTURAL**

**INSURANCE**

**ACCESS**

Medical care

Financial burden on family

Poor outcomes

- Later stage of dx
- Higher mortality

Medical care
Call to Action

There are currently no large-scale programs in the US that screen for LS in unaffected individuals.

CDC and WHO criteria for population screening for genetic predisposition include:

- Disease is an important public health burden 1:300 individuals have LS
- Risk for disease is known: >80% CRC risk; Risk known for other cancers
- Effective interventions: CRC screening; prophylactic surgery; cascade testing
DUAL: Detecting UnAffected Individuals with Lynch syndrome

Cancer Prevention Research Institute of Texas

Identification
- General population screening
- Awareness campaigns
- Community outreach
- Cascade testing

Genetic Services
- Navigation of high-risk patients to genetic counseling
- Genetic counseling: telemedicine/in-person
- Genetic testing via at-home saliva kits

Follow Up
- Patient/provider education: management recommendations
- Genetic Patient Navigator: coordinate care + cascade testing
- Assess compliance

CANCER PREVENTION
Multiple Approaches

General Population
- UTSW Mammo/GI
- PHHS Mammo/GI
- CSPAN coalition
- Self referrals
- Physician referrals

Awareness Campaigns
- Family history cards
- DUAL website
- 24 Hour Fitness Campaign

Community Outreach
- Lynch Leadership
- Presentations/publications

Identification
- Family letters
- Improved access via phone counseling
- Testing cost covered

Cascade testing
Navigating Barriers to Genetics Services

**RURAL GEOGRAPHIC ISOLATION**
- Partnership with CSPAN: 23 counties
- Provider education/outreach

**INSURANCE SOCIOECONOMIC STATUS**
- Safety-net hospitals
- Grant funding for uninsured/underinsured

**ACCESS**
- Navigation
- Tele counseling
- Saliva kits

**EDUCATION**
- Awareness campaigns
- Educational handouts
- Community outreach
Genetic Testing

- Panel testing covered by DUAL grant
- Common Hereditary Cancer Panel

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- Saliva samples for at-home testing
Reach within Texas

- 59 counties touched
- 12.6 million impressions via digital signage
  - 24 Hour Fitness digital signage
- 283,269 people reached
- 911 people directly educated
- 1,594 providers reached
- 1,358 providers directly educated
- ~60% of our patients are underserved
Results—30 months

127,938 screened

5,868 (4.6%) high risk

975 underwent GC

855 (88%) elected GT

UTSW/PHHS GI

UTSW/PHHS Mammo

CSPAN

Result available

Pending

Cancelled

No sample

96

134

614
Results Breakdown

- **Positive**: 335 (55%)
- **VUS**: 202 (33%)
- **Negative**: 77 (12%)
Mutation Spectrum

- **31%**
  - ATM
  - HOXB13
  - RECQL4
  - BARD1
  - PALB2
  - CDKN2A
  - RAD51C
  - FH
  - RAD51D

- **29%**
  - BRCA2
  - NF1
  - BRIP1
  - NBN
  - CHEK2
  - PALB2
  - AXIN2
  - MSH6/MUTYH (het)

- **40%**
  - BRIP1
  - APC
  - NTHL1 (het)
  - MLH1
  - MTHY (het)
  - MSH2
  - MSH6
  - PMS2

16 (52%) unaffected by CRC
Challenges

• “No show” rate: 32%
• Tests not completed: 11%
• Cascade Testing: <1:1 (Goal = 1:2)

Next Steps

• Automation of EMR family history screening
• Automated messaging for colonoscopies
• Improve uptake of phone counseling, test completion, cascade testing
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