CASE STUDY

Delayed colon cancer diagnosis results in death

CASE SUMMARY

A 37-year-old man presented after experiencing poor appetite and jaundice for the past two weeks. The patient was uninsured, related to income issues and complications with the Affordable Care Act (ACA) insurance website. He couldn't afford private insurance but his income was too high to qualify for ACA subsidies.

When he nonetheless tried at the last minute to enroll in the health exchange online, he was unable due to crashing computers, but was told that the State recognized the issue and to apply the following week after the deadline. However, when he tried again he was told he had missed the deadline.

Thus, the patient did not have insurance or a primary care provider when he began to experience malaise, poor appetite, and yellowing of his eyes. He consulted a physician friend who made a presumptive diagnosis of hepatitis- most likely hepatitis A due to lack of risk factors for other types of hepatitis.

A parsimonious set of labs were ordered because they were to be paid out-of-pocket: liver function tests and hepatitis A IgM. Liver enzymes returned showing high bilirubin (14), mildly elevated liver enzymes, and negative hepatitis A IgM. The lab failed to include an ordered alkaline phosphatase in the liver screen.

The patient at this point also mentioned that he had been having rectal bleeding for the past eight months, which he had assumed was due to his hemorrhoids, and that he had a positive family history of colon cancer, with his father dying from colon cancer at age 38. The patient had never been screened for colon cancer despite having been enrolled in a managed care plan for a 2-year period, five years prior, and he was not aware of any guidelines regarding earlier screening.

A repeat bilirubin (20) and alkaline phosphatase (850) suggested worsening jaundice likely due to biliary obstruction. A CT scan of the abdomen was ordered ($1,000 out-of-pocket costs) and showed a large (8cm) left lower quadrant irregular mass suggestive for sigmoid colon cancer, a 5cm liver mass compressing the biliary system, and multiple enlarged abdominal lymph nodes. A subsequent colonoscopy showed a fungating, ulcerated, partially obstructing mass in the proximal rectum extending into the sigmoid colon. Pathology was positive for invasive adenocarcinoma.

The patient underwent multiple biliary stents, abdominal surgeries and rounds of chemotherapy for metastatic colon cancer but ultimately died at age 41. Genetic testing for various known "inheritable" colon cancers (e.g. Lynch syndrome, familial adenomatous, polyposis) was negative.

ISSUES IDENTIFIED

1. No health insurance

Income and cost issues:

- Self-employed with limited income, unable to afford private health insurance
- Income too high to qualify for ACA subsidies
- High out-of-pocket costs for tests and imaging

Technical issues with ACA website:

- Misinformed: patient was told he could enroll after deadline due to server issues but then informed that he missed the deadline and could not enroll

2. Other insurance issues

- Coverage for "screening" colonoscopy (no co-pays) doesn't apply if patient found to have polyp or if done to follow-up positive stool test for blood
- After being referred and scheduled, unable to access hospital care owing to lack of insurance coverage
- Covered by various health insurance plans over prior decade, including a highly-regarded managed care plan, yet there were discontinuities and patient was never offered screening
- Patient not directed to free care options

CHANGE IDEA

- CMS and Medicare (under the ACA) consider biopsy and polypectomy integral to diagnostic but not screening exams. This does not automatically apply to Medicaid or private carriers, but most have adopted the same definitions. Legislative remedies are needed to remedy this.

3. Problems with obtaining and updating family histories in primary care

- Guidelines known to physicians but not to patients
- Discontinuities between deceased father's care three decades earlier and present
- Potential privacy concerns in sharing family history

CHANGE IDEA

- Need to systemically inform family members of heightened risk and the need for early or more intensive screening. Potential for linking medical records of family members to trigger screening reminders and/or family history documentation.

4. Problems with obtaining and updating family histories in primary care

- Unclear whose job it is to obtain and update family history, how often and when
- Adopted individuals and new immigrants often do not know family history or what kind of cancer their relatives had
- Confusion regarding correlation between first-degree and second-degree relatives
- Confusion about results, response to genetic test a relative had received
- Uncertainty about whether relative had cancer versus a polyp, and how best to schedule screening based on this
- Role of genetic testing for Lynch syndrome in patients who have had relatives with uterine cancer

5. Patient failure to report rectal bleeding

- Assumed was due to prior history of hemorrhoids

6. Failure of prior managed care provider to perform recommended cancer screening

- Should have been offered a screening at 28 if he had been carefully tracked by primary care provider. If he declined the colonoscopy, he should have been offered a FIT test and annual testing as an alternative

7. Genetic testing: Cost, negative result

- Insurance refused to cover genetic testing despite patient's diagnosis and positive family history
- Patient had likely genetic risk for colon cancer, yet when finally tested, no detectable known abnormality

TAXONOMIES

Diagnosis Error Evaluation and Research (DEER) Taxonomy

Where in the diagnostic process an error may have occurred

1. Access/Presentation	a. Failure/delay in presentation b. Failure/denied care access
2. History	a. Failure/delay in eliciting critical piece of history data b. Inaccurate/misinterpreted/overlooked critical piece of history data c. Failure in weighing critical piece of history data d. Failure/delay to follow-up critical piece of history data
3. Physical Exam	a. Failure/delay in eliciting critical physical exam finding b. Inaccurate/misinterpreted/overlooked critical physical exam finding c. Failure in weighing critical physical exam finding d. Failure/delay to follow-up critical physical exam finding
4. Tests (Lab/Radiology)	*Ordering (also called "pre-analytic phase")* a. Failure/delay in ordering needed test(s) b. Failure/delay in performing ordered test(s) c. Error in test sequencing d. Ordering of wrong test(s) e. Tests ordered to be done in the wrong way *Performance (also called "analytic phase")* f. Sample mix-up/mislabeled (e.g., wrong patient/test) g. Specimen delivery problem h. Technical errors/poor processing of specimen/test i. Erroneous lab/radiology reading of test j. Failed/delayed reporting of result to clinician *Clinician Processing (also called "post-analytic phase")* k. Failed/delayed follow-up of (abnormal) test result l. Error in clinician interpretation of test
5. Assessment	*Hypothesis Generation* a. Failure/delay in considering the diagnosis *Suboptimal weighing/prioritizing* b. Too little consideration/weight given to the diagnosis c. Too much weight on competing/coexisting diagnosis *Recognizing urgency/complications* d. Failure/delay to recognize/weigh urgency e. Failure/delay to recognize/weigh complications of a diagnosis
6. Referral/Consultation	a. Failure/delay in ordering referral/consult b. Failure/delay in obtaining/scheduling ordered referral c. Error/suboptimal quality in diagnostic consultation performance d. Failed/delayed communication/follow-up of consultation
7. Follow-up	a. Failure/delay in timely follow-up/rechecking of patient b. Failure to refer patient to close/safe setting/monitoring c. Failure/delay in needed monitoring or lab (BP, INR, repeat CXR) d. Failure/delay in communicating findings among healthcare providers

Reliable Diagnosis Challenges (RDC) Taxonomy

Factors that may have contributed to making diagnosis difficult

1. Challenging Disease Presentation	a. Rare diagnosis b. Atypical presentation c. Nonspecific signs and symptoms d. Unfamiliar/outside specialty e. Masking/mimicking diagnosis f. Red herring misleading finding (history, exam, lab/imaging) g. Rapidly progressive h. Slowly evolving i. Deceptively benign (or intermittent) course
2. Patient Factors	a. Language/communication b. Signal: noise (noisy pts with multiple nonspecific sx) c. Patient failure to share d. Patient failure to follow-up
3. Testing Challenges	a. Test availability, access, cost b. Logistical issues in obtaining, performing tests c. False positive/negative results d. Performance/interpretation challenges e. Equivocal results/reports f. Test follow-up issues
4. Stressors	a. Time constraints b. Discontinuities c. Fragmentation of care d. Memory reliance/challenges e. EMR challenges
5. Broader Challenges/ Failures	a. Recognition of acuity/urgency/severity b. Diagnosis of complication(s) c. Recognizing failure to respond to treatment d. Diagnosis of underlying cause e. Recognizing misdiagnosis

Generic Diagnostic Pitfalls Categories

Clinical patterns/vulnerabilities leading to missed, delayed or wrong diagnosis

1. Diagnosis/ Assessment	a. Disease A misdiagnosed/confused with Disease B b. Misled by atypical presentation c. Rare diagnosis: failure to consider or know d. Chronic disease presumed to account for new symptoms (especially in medically complex patients) e. Counter-diagnosis cues overlooked (e.g., red flags, things that don't fit not recognized) f. Drug or environmental factor overlooked as cause of symptoms, or as cause of disease progression g. No specific diagnosis made
2. History/ Physical	a. Non-specific/vague symptom(s); hard-to-pinpoint diagnosis b. Intermittent symptoms- overlooked because findings (e.g., exam, lab, EKG) negative when seen c. Failure to appreciate risk factor (or those at risk) for a given disease d. Failure to appreciate limitations of the physical exam
3. Testing	a. Failure to appreciate limitations of a test result(s) b. Failure in follow-up of abnormal/critical result
4. Communication	a. Communication failure with patient, including language barriers b. Failure around communication and ordering of lab tests c. Communication failure between physicians (e.g., PCP-specialist, ED-PCP)
5. Follow-up	a. Failure to monitor, note, or respond to evolving/continuing/persistent symptoms b. Inadequate follow-up visits/referrals, especially in the presence of diagnostic uncertainty
6. Other	a. Urgency of the clinical situation was not appreciated b. Diagnostic findings were masked or misinterpreted due to an intervention or drug (e.g., empiric treatment with oral or topical steroids, PPI, antibiotics, pain medications) c. Problems with inappropriate or over-referral

Cognitive Errors Taxonomy

Selected cognitive biases contributing to diagnostic errors

1. Premature Closure: accepting a diagnosis before it has been fully verified
2. Anchoring: tendency to fixate on specific features of a presentation too early in the diagnostic process and subsequent failure to adjust
3. Confirmation Bias: tendency to look for confirming evidence to support one's hypothesis, rather than disconfirming evidence to refute it
4. Search Satisfying: tendency to call off a search once a piece of data is found, and not considering/searching for additional findings or diagnoses
5. Availability Bias: tendency to give too much weight to diagnosis that come more readily to mind (e.g. recent dramatic case).
6. Base-Rate Neglect: failing to adequately take into account the prevalence of a particular disease
7. Knowledge Deficit (on part of provider)
8. Demographic/Stereotype Bias: Biases from personal or cultural beliefs about women, minorities or other patient groups for whom prejudices may distort diagnostic assessment
9. Other (please specify)

Primary-Care Research in Diagnosis Errors (PRIDE) is an effort to improve diagnostic safety, led by Brigham and Women's Center for Patient Safety Research and Practice in partnership with Gordon and Betty Moore Foundation and the Betsy Lehman Center.

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