CASE REPORT

Diagnostic error: what Muir-Torre syndrome has taught us

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SUMMARY

A low-grade sebaceous carcinoma was excised from a 55-year-old woman's neck. At follow-up, 11 months later, a recent diagnosis of mucinous adenocarcinoma of the colon and history of clear cell endometrial carcinoma were identified. A strong family history of bowel cancer suggested Muir-Torre syndrome. Unexpectedly, hereditary non-polyposis colorectal cancer had previously been genetically confirmed; the results were found loosely filed in the paper chart. The patient had not informed us about her diagnosis; having discussed the case with other physicians she felt the diagnosis would be common knowledge. In 1999, US National Institute of Medicine estimated that preventable medical error resulted in the deaths of 44–98 000 people yearly in US hospitals. Four categories of medical error, including diagnosis, treatment, preventive and 'other' were described, while the reasoning processes that result in these errors are outlined by Reason *et al.* We utilise this rare case to illustrate these important concepts.

BACKGROUND

Physicians are human and as such are prone to error. Unfortunately, the traditional 'person approach' is commonly adhered to in medical practice, whereby a person is sought out to blame when errors occur.¹ The expanding medical error literature reveals how high reliability organisations utilise an alternative approach to significantly reduce errors, the 'system approach'. In this model, the fallibility of humans is accepted and the emphasis is on optimising the systems in which they work to help reduce errors.

As a physician I have been concerned about how my actions might harm patients. Discovering the field of medical error has been significant for me in that it has helped me become aware of situations that can lead to preventable errors and understand how I might avoid them. It was an area that was not highlighted during my training, but one that I feel that all physicians should have the opportunity to engage with to help us shift from a community who adhere to the 'person approach' to the 'system approach'. It is also a system that facilitates the inclusion of evidence-based medicine and technology that can facilitate the safe practice of medicine which will ultimately benefit both the patient and our own health.

This case is one that has helped me recall an uncommon but very significant syndrome, while also strikingly illustrating and facilitating recall of many of the concepts that result in preventable medical error.

CASE PRESENTATION

A 55-year-old woman presented with an 8-week history of a gradually enlarging lesion on the neck (*figure 1*). Medical history included uterine cancer in 2007 which was treated with hysterectomy, radiotherapy and chemotherapy. No family history of skin cancer was noted. A recent diagnosis of diabetes was revealed during serial attendances in the context of a postoperative wound infection. Histology demonstrated an ulcerated, markedly inflamed tumour, composed of lobules of sebocytes without surrounding desmoplasia and prominent mitoses; this raised suspicions about a diagnosis of a *low-grade sebaceous carcinoma* (*figure 2A, B*). The report, discussed further at a dermatopathology meeting, suggested that extraocular sebaceous carcinomas tend not to be associated with underlying malignancy but may recur locally. At a follow-up clinic, 11 months postsurgery, it emerged that a series of reviews by other medical specialties had recently culminated in a right hemicolectomy for excision of a bowel mass. Family history revealed multiple cases positive for bowel cancer, and the non-linked digital pathology records identified a history of rare *clear cell endometrial carcinoma* and *mucinous adenocarcinoma* of the bowel. Loosely placed among other numerous, random reports in the patient's chart was a genetics report, performed by the Oncology services posthysterectomy, noting a c.942+3A>T splice mutation in IVS5 of the MSH2 gene. This confirmed our suspicion of hereditary non-polyposis colorectal cancer (HNPCC) and therefore, Muir-Torre syndrome (MTS). Although the patient had discussed this diagnosis with a number of other physicians in the past, she had not disclosed this to us as she assumed that all

Figure 1 An exophytic, crusted nodule on the left side of the neck.

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Figure 2 (A) An ulcerated (circled), markedly inflamed tumour, composed of lobules of sebocytes (arrow) without surrounding desmoplasia. (B) Higher power showing prominent mitoses and a histological appearance highly suspicious of low-grade sebaceous carcinoma.

physicians would be aware of her diagnosis. It was revealed that with the exception of regular gynaecological review, no other screening of the patient had occurred.

OUTCOME AND FOLLOW-UP

Fortunately, 41 months postexcision of the sebaceous carcinoma from the neck, 31 months postexcision of the bowel mucinous adenocarcinoma, 7 years postendometrial carcinoma and 6½ years postgenetic diagnosis, there has been no evidence of malignancy despite ongoing surveillance by multiple specialities.

DISCUSSION

Muir-Torre is an autosomal dominant syndrome with incomplete penetrance. It is associated with an inherited defect in one copy of a DNA mismatch repair genes MLH1, MSH2 (mutS homolog2), MSH6 or PMS2 (postmeiotic segregation).² It is considered to be a phenotypic variation of HNPCC, synonym: Lynch syndrome and is the association of sebaceous gland tumours or keratoacanthomas with one or more visceral cancers.^{2 3}

A review of the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) database reveals that the specific mutation identified in this case (c.942+3A>T in IVS5 of the MSH2 gene) has been reported on 168 prior occasions.⁴ The literature suggests that mutations in MTS are most commonly identified in MSH2.⁵ Of note, a specific study—focused on detecting the frequency of MTS in families with Lynch syndrome—identified that carrying the specific c.942+3A>T MSH2 mutation had a significantly higher frequency of MTS compared with families with other MSH2 gene mutations.⁶

In 1999, the US National Institute of Medicine published an article entitled ‘To Err is Human’.⁷ They noted that between 44 000 and 98 000 people die due to preventable medical error in hospitals in the USA annually, based on the estimates from two major studies. They referred to Leape *et al*⁸ in identifying four major types of error relating to: diagnosis, treatment, preventive and ‘other’.⁷ Each category has further subclassifications (figure 3). This case highlights a number of errors:

Diagnostic

1. An error or delay in making a diagnosis
2. A failure to act on results of monitoring or testing

The other two types of errors in this category are:

- Failure to employ indicated tests and

Figure 3 The four major types of error types and subclassifications, with the errors demonstrated in this case highlighted in red.^{7 8}

- ▶ The use of outmoded tests or therapy

Treatment

3. Avoidable delay in treatment or in responding to an abnormal test

Other errors in this group include:

- ▶ Error in the performance of an operation, procedure or test
- ▶ Error in administering the treatment
- ▶ Error in the dose or method of using a drug
- ▶ Inappropriate (not indicated) care.

Preventive

4. Failure to provide prophylactic treatment
5. Inadequate monitoring or follow-up treatment

Other

6. Failure of communication in that the patient should have been referred for routine colonoscopy when HNPCC was initially diagnosed

Other errors include equipment failure and 'other system failure'.

Diagnostic error can also be considered with respect to the reasoning process behind the errors. A number of authors have suggested a terminology for describing unsafe acts (figure 4).^{1 9 10}

Slips

A *slip* is a deviation from the correct plan; in this case, a slip occurred due to overlooking the association between sebaceous carcinoma and MTS, perhaps due to the patient and her case being viewed out of context, such as in the wound review clinic or at the dermatopathology meeting, rather than at the formal outpatient review or other coexisting problems diverting attention, such as the wound infection and her concerns about it.

Lapses

A *lapse* is an unintended deviation from the correct plan due to memory failure; in this case, outpatient follow-up was not given to the patient from oncology following her ovarian carcinoma.

Mistakes

A *mistake* is when the planned action to reach a certain goal deviates from the action required to reach the goal; in this case, the surgical team's error was not knowing that the patient's particular type of bowel cancer was associated with sebaceous carcinoma.

Record Error

This is where the correct act is performed, but not noted in the patient record; in this instance, genetic testing reports were loosely placed randomly with numerous other items in the patient's chart. Since this presented case, new medical records in our institution are now less prone to this specific error as they contain specific sections delineated for filing results and thus, making them more observable.

Violations

A *violation* is a deliberate deviation from some regulated code of practice or procedure.

Figure 4 Identifies unsafe acts that may result in medical error with those encountered in this case highlighted in red.^{1 9 10}

Two suggested approaches are described in the literature with respect to preventing medical error.¹ The first is the traditional 'person approach' where unsafe acts arise from aberrant mental acts; someone is to blame. The 'system approach' is that taken by high reliability organisations, such as the nuclear power industry, where failures have harmful consequences.¹ It accepts that humans are fallible and rather than trying to ascribe blame, the system in which they work is optimised to help prevent errors by reducing reliance on memory, improving information access, error proofing, standardisation and training.¹⁰

We encourage the system approach. This is a similar concept to that of performing audit: discover your weaknesses and provide mechanisms to help avoid them. We are drawing attention to this case as a good teaching example of the multiple errors that can occur, despite a patient progressing through numerous competent physicians and specialties. We feel that it is important to discuss similar cases in the literature as

Patient's perspective

I am very happy to have my case published as I feel that it might:

- ▶ Highlight this rare condition and raise awareness of associated cancers.
- ▶ Highlight the difficulty that exists with making investigation results and full medical history available to all physicians, across practices, with a view to overcoming this problem.
- ▶ Increase awareness of preventable medical error and the factors that can lead to its occurrence.
- ▶ Give me the opportunity to suggest that there should be a mechanism of assuring all physicians are fully aware of the information that is in the patient file or have important results highlighted in a separate section of the patient file that all physicians would have to read when first meeting that patient.

I note that, subsequent to my case, hospital notes have since been introduced to my hospital that do not have a sleeve where loose sheets may be filed. All notes must now be inserted, in appropriate sections to avoid notes falling out. A summary sheet placed on the front of the paper notes now exists, however this still relies on individuals listing significant diagnoses. I am hopeful that solutions, possibly electronic, will emerge that solve the difficulty of logically presenting a succinct summary of a patient with a complex history, which would be of particular relevance for cases such as mine.

Learning points

- ▶ Muir-Torre syndrome (MTS) is the association of sebaceous gland tumours or keratoacanthomas, with one or more visceral cancers.
- ▶ MTS is an autosomal dominant syndrome with incomplete penetrance and is a phenotypic variation of hereditary non-polyposis colorectal cancer.
- ▶ Medical error can be analysed with a 'person approach' where someone is to blame or the 'system approach' utilised by high-risk organisations, such as nuclear power plants, whereby people are accepted as being fallible and the systems in which they work are optimised to reduce error.²

proponents of the system approach, noting that we will not avoid the errors we strive to prevent occurring if we do not first acknowledge them openly.

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