CASE REPORT

Sister Mary Joseph nodule: a diagnostic challenge

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SUMMARY

We present the case of a 72-year-old woman who presented with, to our knowledge, the largest reported Sister Mary Joseph lesion in the literature. Often associated with a poor prognosis, the patient went on to have a wide local excision of the lesion and has made a full recovery 2 years after the nodule initially developed. Histological examination confirmed the presence of underlying endometrial cancer and the patient subsequently underwent a total hysterectomy and bilateral salpingo-oophorectomy.

BACKGROUND

First described in 1854,¹ the Sister Mary Joseph nodule refers to an umbilical metastasis from internal malignancy. Clinicians must be aware of the consequences of discovery as the nodule is often the presenting symptom of new malignancy or cancer recurrence.^{2–4} The case presented here demonstrates the power anxiety has over patient behaviour. Despite being the largest reported case in the literature, identification of primary malignancy was difficult, requiring extensive investigation. The prognosis of the patient has exceeded the mean survival time, supporting the notion of surgical treatment in patients with no identifiable metastases.

CASE PRESENTATION

A 72-year-old woman attended her general practitioner for her annual influenza vaccination. It was noted that she was mobilising with extreme difficulty and was profoundly dyspnoeic. On further questioning, she reported a 1-year history of progressive fatigue. On systemic examination, she was remarkably pale. Examination of the cardiorespiratory system was unremarkable. On abdominal examination, a 180×120×60 mm fungating mass arising from her anterior abdominal wall was identified (figures 1 and 2). On further questioning, the lesion had been present for a total of 18 months and been episodically bleeding. Her medical history included hypertension, for which she was taking treatment. The patient lived alone and despite having a son, she had not informed anyone of the tumour. The patient stated extreme anxiety and fear of healthcare professionals for her late presentation. She stated that she attempted to live with the nodule.

INVESTIGATIONS

Routine laboratory investigations confirmed iron deficiency anaemia, with a haemoglobin of 78 g/L. Her inflammatory markers were also raised at this time. Of note, her tumour markers including Ca

19–9, CA 125 and Carcinoembryonic antigen were within normal limits.

She received two units of packed red cells and underwent a CT scan of her abdomen which further defined the lesion as being confined to the abdominal wall and normal intra-abdominal organs. A wedge biopsy identified the lesion as a secondary umbilical tumour or Sister Mary Joseph nodule, most likely arising from an ovary of gynaecological organ owing to high expression of progesterone and oestrogen receptors. The lesion was sent to an additional pathologist for a second opinion. Who concluded that the lesion was an adenocarcinoma of unknown origin.

She was discussed at the local multidisciplinary team meeting which recommended further radiological staging prior to excision. An MRI scan of the pelvis did not provide any further information on the primary site of malignancy; the pelvic organs were unremarkable. A positron emission tomography scan did not highlight any further metastases (figure 3).

Histological examination of the excised lesion, confirmed the results of the wedge biopsy; the tumour was a grade 3 endometrioid adenocarcinoma with squamous differentiation. The tumour was oestrogen receptor (ER) and progesterone Receptor (PR) positive.

The presence of the oestrogen and progesterone receptors suggested a primary malignancy in the gynaecological organs.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis of the lesion in question includes a primary umbilical malignancy or secondary umbilical malignancy. With regards to the secondary malignancies, the most common primary source of the lesion in women is from the gynaecological tract. In men, the gastrointestinal tract and pancreatobiliary tract are the most frequent.² While initially reported by Sister Mary Joseph as arising from an intra-abdominal source, cases have been reported from the lung,⁵ breast and lymphoma.⁶

TREATMENT

The lesion was excised with the assistance of plastic surgeons (figure 4). The deep margins of the lesion were superficial to the anterior sheath and with a 3 cm lateral margin, the wound was closed primarily.

OUTCOME AND FOLLOW-UP

Her postoperative recovery was hampered with an acute kidney injury; however, this resolved

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Figure 1 The lateral aspect.

with conservative measures. Her wound healed without complications.

The findings of the aforementioned investigations were discussed with the gynaecological multidisciplinary team meeting and the decision was made to perform a total hysterectomy and salpingo-oophorectomy.

Histological examination of the uterus, fallopian tubes and ovaries identified synchronous type 1 endometrioid endometrial adenocarcinoma and a grade 2 endometrioid adenocarcinoma within the ovary. The immunohistochemical profile of the ovarian lesion was similar to that of the umbilical tumour.

The patient recovered uneventfully from this procedure. A repeat CT scan identified no further malignancy. She was then referred to oncology for consideration of chemotherapy. The patient declined chemotherapy and was offered hormone therapy. The patient is currently taking the aromatase inhibitor letrozole.

DISCUSSION

The Sister Mary Joseph nodule refers to an umbilical, cutaneous metastases of an internal malignancy. The lesion was first described in 1854 by Baluff¹; however, the eponym was first described by Hamilton Bailey in the 11th edition of 'Physical Signs in Clinical Surgery' in acknowledgement of Sister Mary Joseph Dempsey, assistant to Dr William James Mayo. Sister Mary Joseph noted a relationship in patients with umbilical nodules whose subsequent surgery identified intra-abdominal malignancy.⁷

Figure 2 The lesion in vivo.

Figure 3 An axial section of MRI and PET scan of the lesion. PET, positron emission tomography.

Classically, the Sister Mary Joseph nodule presents as a firm, irregular nodule that is often ulcerated and painful. Typically, it is below 5 cm in diameter. In some cases, a diffuse induration of subcutaneous tissue has been reported instead of a specific nodule.⁸ It is more common in women. Recognition of the lesion is essential as it is often the first presentation of internal malignancy or of cancer recurrence.⁹

Differential diagnosis of an umbilical nodule includes an umbilical hernia, primary umbilical malignancy, benign lesions such as keloid scars and endometriosis. In a review of 77 patients presenting with an umbilical lesion, metastatic secondary cancers were more common than primary.¹⁰

Adenocarcinoma is the most frequent histopathological appearance. Squamous cell carcinoma, mesothelioma, leiomyosarcoma and cholangiocarcinoma have all been reported.² The most common primary source of the lesion in women is from the gynaecological tract. In men, the gastrointestinal tract and pancreatobiliary tract are the most frequent.² While initially reported by Sister Mary Joseph as arising from an intra-abdominal source, cases have been reported from the lung,⁵ breast and lymphoma.⁶ As with the case reported, it is important to highlight that identifying a primary site may be challenging and in some cases, not identified.⁹

There are a variety of routes in which malignancies can spread to the umbilicus. The embryological formation of the umbilicus creates an extensive relationship with the lymphatic and vascular systems. In addition, direct spread can occur from the peritoneum and falciform ligament.¹¹

Figure 4 The excised lesion.

Sister Mary Joseph nodules often represent advanced malignancy and are typically associated with a poor prognosis. The mean survival from diagnosis is 11 months.² Owing to the poor prognosis, treatment is often palliative aimed at symptom control. Confined disease and unknown primary source of cancer are associated with a better prognosis, in such cases excision is warranted. The longest survival reported in the literature is 18 years from diagnosis.²

Learning points

- ▶ The Sister Mary Joseph nodule is often the presenting symptom of underlying malignancy or cancer recurrence.
- ▶ In some instances, identifying the primary malignancy can be difficult and requires a multidisciplinary approach.
- ▶ Sister Mary Joseph nodules often represent advanced malignancy and are typically associated with a poor prognosis.

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