Simultaneous Multiple Pulmonary Emboli in Monozygotic Twins with Dercum’s Disease: A Timely Coincidence?

J. Raikes, I. Weichert

Department of Acute Medicine, The Ipswich Hospital, Ipswich, Suffolk, United Kingdom

INTRODUCTION

Several genetic determinants (mutations or polymorphisms) have been associated with increased risk of venous thromboembolism but the overall influence of genetic factors on this disease is unknown. A retrospective study by Larsson et al in 2003 demonstrated an increase in the relative risk of venous thromboembolism in male monozygotic twins only. As yet there is no published evidence which supports an intra-twin pair similarity for women.

Dercum disease (Adiposis Dolorosa) is a rare condition characterised by multiple, painful lipomas. The lipomas are typically found on the trunk, upper arms and upper legs. The underlying pathology of the disease is poorly understood, but as yet there has been no recorded association with an increased risk of venous thromboembolic disease.

OBJECTIVES

To report a case of two female monozygotic twins, both suffering from Dercum’s disease, presenting at the same time with multiple pulmonary emboli.

CASE REPORT

TWIN 1

A 67-year-old woman with a past medical history of Dercum’s disease and Rheumatic fever in childhood presented with a four-month history of shortness of breath on exertion with no chest pain and no symptoms at rest. Multiple lipomas were detected consistent with her underlying chronic condition, but cardiovascular, respiratory, gastrointestinal and neurological examinations were otherwise normal. CXR was normal. D-Dimer was elevated at 3100 and VQ SPECT scanning revealed multiple pulmonary emboli (image 1). As per local guidelines, Warfarin was commenced with sub-cutaneous Enoxaparin cover at 1.5mg/kg of bodyweight until the INR was therapeutic for two days. Outpatient investigations looking to identify an underlying cause were unyielding (see table I for summary of results).

TWIN 2

Three weeks later, the patient’s monozygotic twin was admitted with a six-month history of gradually worsening shortness of breath on exertion. The recent admission of her twin sister with symptoms which mirrored her own had prompted the patient to seek medical advice. The patient was otherwise well but also had a diagnosis of Dercum’s disease. Aside from multiple lipomas in line with her past medical history, general examination was unremarkable. Again, CXR was unremarkable and D-Dimer was elevated. VQ SPECT scanning demonstrated multiple pulmonary emboli (image 2). Warfarin was commenced with Enoxaparin cover until the INR was therapeutic for two days. Further investigations were performed as an outpatient (see table).

The patient herself had identified a lipoma in the groin area and had raised the suspicion of a pressure effect on the underlying vasculature in this area. In an ultrasound scan the abdominal and pelvic organs appeared normal with no evidence of sinister mass lesions at any site. No mass lesions were seen in the right groin at the site indicated by the patient. The right femoral vein appeared normal.

DISCUSSION

The overall influence of genetic factors on VTE remains unknown. A retrospective study by Larsson et al in 2003 used the Danish Twin Registry and Danish National registry of patients to access all hospitalisations in Denmark since 1977. This demonstrated that 678 twins were hospitalised with an episode of VTE. The odds ratio was 1.53 at the relative risk of VTE for one twin, given VTE in the partner twin was 13.5 among monozygotic twins and 3.8 among dizygotic twins. Interestingly they found differences in genetic susceptibility to venous thromboembolism between the sexes, with genetic factors playing a substantially stronger role in males than in females. The study demonstrated an increase in the relative risk of venous thromboembolism in male monozygotic twins but no intra-twin pair similarity for was observed in women.

CONCLUSION

As yet there is no evidence to support an increased risk of VTE amongst female twins. Thus, a set of female twins presenting at the same time, with the same condition is extremely rare. Attempts were made to uncover a unifying, underlying diagnosis that could have predisposed these two women to developing VTE disease. Blood tests undertaken to investigate the possibility of predisposition towards thrombophilia were negative. Furthermore, tumour markers, which might point towards an underlying malignancy causing a hypercoagulable state, were not raised.

The twins have a shared medical history of Dercum’s disease (adiposis dolorosa), which is a rare disorder resulting in painful fatty deposits around the upper legs, trunk, and upper arms. Little is known about the pathological mechanisms of this condition, although it is suspected that there is either a metabolic or autoimmune component involved. We questioned whether the multiple lipomas seen in Dercum’s disease could increase the likelihood of DVT formation via a direct mechanical effect causing deep venous compression. However, in this case a scan of the abdomen and pelvis was performed which detected no evidence of venous compression. Review of the literature revealed that as yet there has been no recorded association with an increased risk of venous thromboembolic disease.

REFERENCES