J Forensic Sci, Sept. 2005, Vol. 50, No. 5
Paper ID JFS2005001
Available online at: www.astm.org

CASE REPORT

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Autopsy Confirmation of Severe Pulmonary Interstitial Fibrosis Secondary to Munchausen Syndrome Presenting as Cystic Fibrosis

ABSTRACT: Chronic factitious disorder with physical symptoms, or Munchausen syndrome, is a well-recognized but uncommonly diagnosed psychiatric condition characterized by the deliberate production of signs and symptoms of disease in order to receive medical attention. Clinical suspicion of this disease is rarely confirmed by autopsy, as the patients usually do not die as a consequence of feigning illness. Here we report the autopsy confirmation of a case of a suspected Munchausen syndrome patient who presented with a history of cystic fibrosis. Examination of the lungs demonstrated extensive severe interstitial fibrosis, and polariscopic examination revealed a large quantity of crystalline material throughout the tissue; X-ray diffraction identified the material as talc. Synopses of published cases of Munchausen syndrome presenting as cystic fibrosis, and cases of Munchausen syndrome with pulmonary talcosis are presented as part of the discussion.

KEYWORDS: forensic science, autopsy, Munchausen syndrome, cystic fibrosis

Chronic factitious disorder with physical symptoms, more commonly known as Munchausen syndrome, is a psychiatric condition in which the patient deliberately produces signs and symptoms of illness for the sole purpose of receiving medical care. Literature on Munchausen syndrome is generally confined to the case report level, where a panoply of presentations has been described. Pulmonary manifestations are unusual in Munchausen syndrome, and cystic fibrosis is a particularly infrequent presenting complaint. Death that is recognized as attributable to Munchausen syndrome is a rare event. In light of this, the following case report describes a patient exceptional even for Munchausen syndrome, for she presented as cystic fibrosis, died from her illness, and underwent a limited yet illuminating autopsy.

Case Report

A 53-year-old white woman with a history of chronic bronchiectasis was hospitalized secondary to worsening respiratory symptoms. The patient, a former nurse, maintained that she was diagnosed with cystic fibrosis in infancy and had been followed in a cystic fibrosis clinic for several years. She had a history of multiple hospitalizations for exacerbations of her lung condition; both medical and surgical interventions were employed during these admissions, including right middle and right lower lobectomies for "pulmonary abscesses," and placement of a transtracheal oxygen catheter.

The patient also had a long psychiatric history for which she had many hospital admissions. She was described as having prominent dissociative symptoms with self-destructive and suicidal tendencies. In addition, she was known to sabotage the efforts of the medical team, and had at one point been diagnosed with Munchausen syndrome.

Although her chest radiographs demonstrated changes consistent with bronchiectasis with extensive scarring, the staff at the cystic fibrosis clinic suspected that the patient did not have cystic fibrosis. She had no known history of a sputum culture positive for *Pseudomonas*, and had repeatedly refused sweat chloride testing. However, the bronchiectasis was due to unknown causes, as the patient had no known occupational or other environmental exposures that could account for this condition. The pulmonologists more recently involved in her care expressed concern that her pulmonary condition was somehow self-induced, a consequence of Munchausen syndrome rather than cystic fibrosis.

During her most recent hospital admission, the patient was treated medically for chronic respiratory failure with pneumonia. Her respiratory function improved, and she was considered sufficiently stable to transfer to a skilled nursing facility. However, while waiting to be transferred, the patient again experienced increased dyspnea, and she was thought clinically to have recurrent bronchitis or pneumonia. After marked deterioration of her condition, she became unresponsive, and subsequently was pronounced dead. An autopsy limited to the chest was performed.

At autopsy, the body was that of a well-nourished, well-developed obese white female. The arms bore numerous stellate scars that had previously been identified clinically as the consequence of self-inflicted injury. Internally, the right and left pleural cavities each contained 10 mL of serosanguinous fluid. The visceral

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Received 8 Jan. 2005; and in revised form 20 April 2005; accepted 23 April 2005; published 3 Aug. 2005.

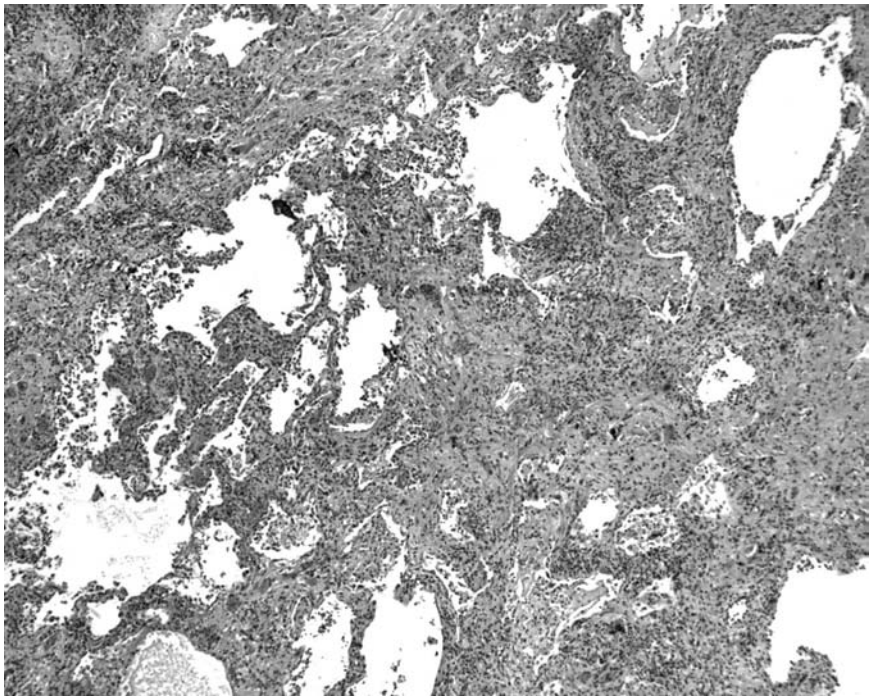

FIG. 1—*Severe interstitial pulmonary fibrosis in a representative lung section, H&E, ×40.*

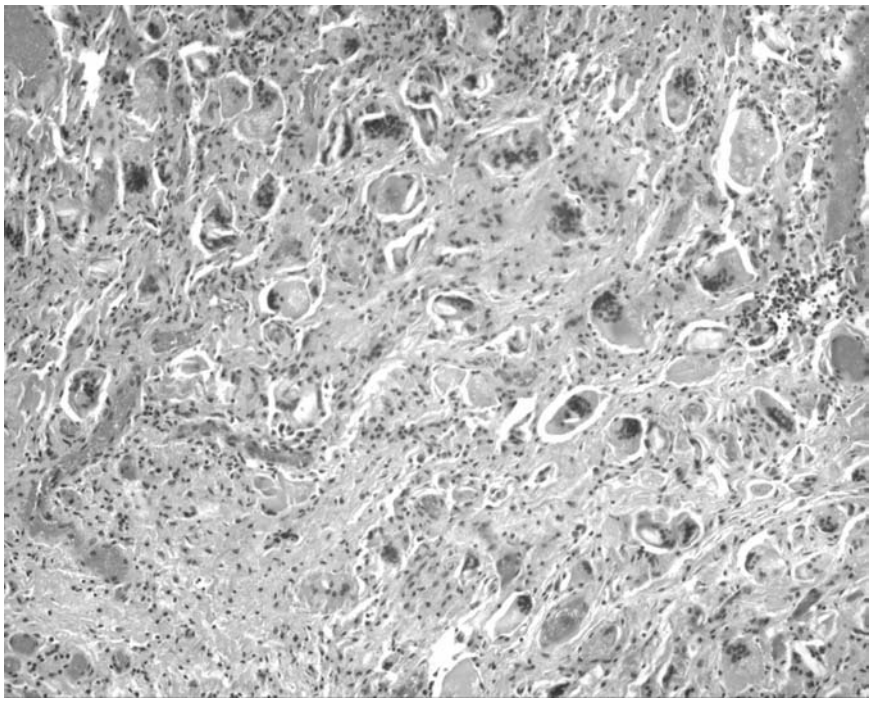

FIG. 2—*Intense foreign-body giant cell reaction, H&E, ×100.*

pleura of both lungs was covered by dense fibrous adhesions which were most extensive inferiorly. The right middle and lower lobes were surgically absent. On cut section, both lungs exhibited multiple peripheral poorly-circumscribed tan nodules, up to 1.5 cm in greatest dimension; the parenchyma was diffusely firm to palpation. Viscous brown fluid was found throughout the respiratory tree, and a 4.5 cm plug of mucus was loosely adherent to the tracheal mucosa. The pulmonary vasculature and hilar lymph nodes were unremarkable. The heart was without significant pathology

Microscopically, all sections of the lungs, including those from the ill-defined peripheral nodules, showed severe interstitial fibrosis with "honeycomb" architecture and an intense foreign-body giant cell reaction (Figs. 1 and 2). Polariscopic examination revealed

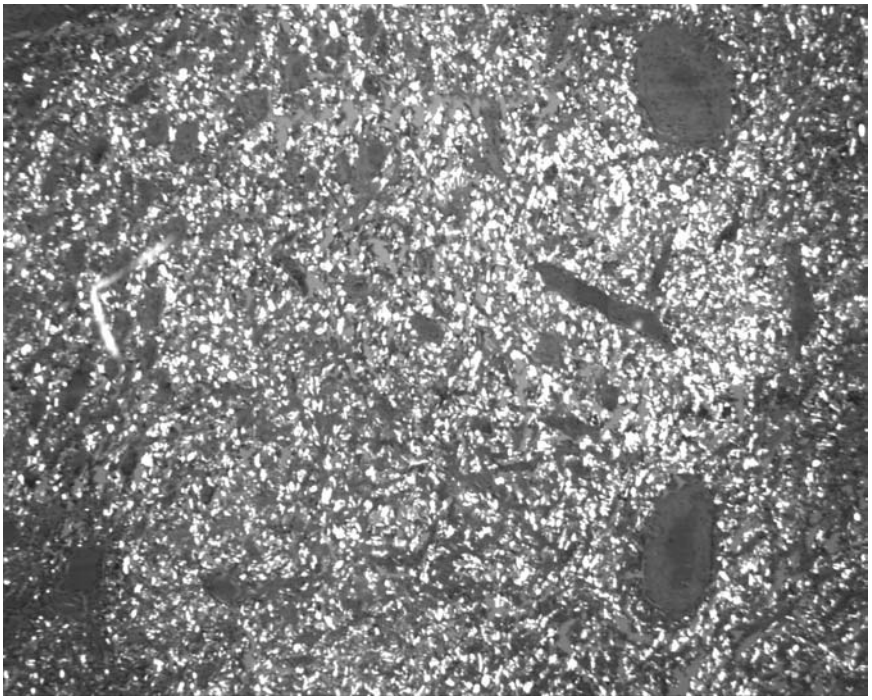

FIG. 3—*Abundant, diffusely distributed birefringent crystalline material in a representative section of lung, H&E, polarized light, ×40.*

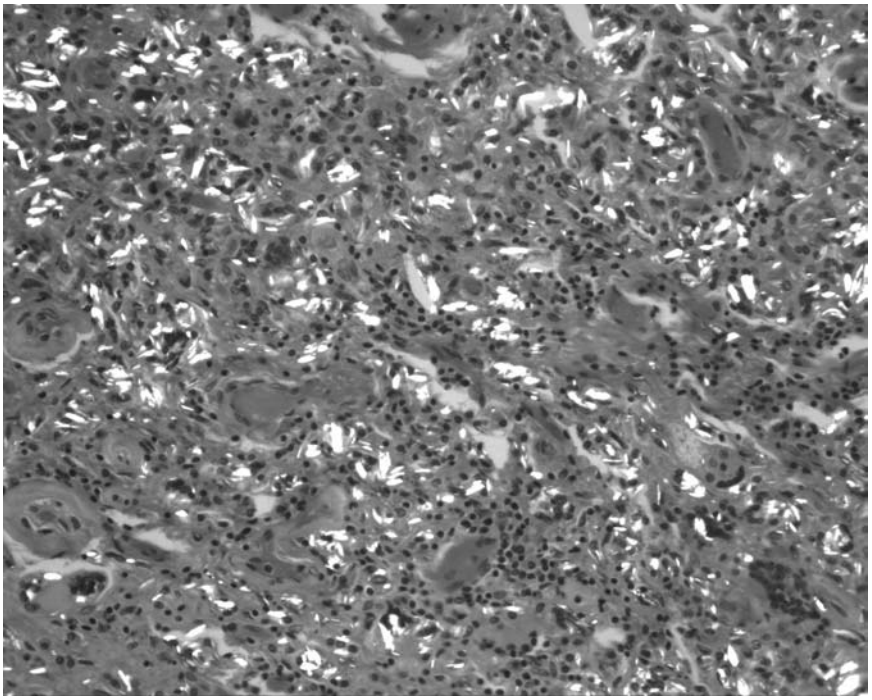

FIG. 4—*Intra- and extra-cellular birefringent crystalline material, H&E, polarized light, ×100.*

very large amounts of intra- and extra-cellular birefringent foreign material with a crystalline appearance throughout the lung sections. (Figs. 3 and 4) This crystalline material was within airspaces and the fibrotic interstitium, and not localized to intravascular or perivascular spaces. A mild bronchopneumonia was also noted, as was focal interstitial osseous metaplasia.

Frozen lung tissue was sent for evaluation of the cystic fibrosis transmembrane regulator (CFTR) gene, looking for the 33 most common cystic fibrosis-associated mutations. The tissue was negative for the mutations analyzed by polymerase chain reaction and oligonucleotide ligation assay.

Death was attributed to severe pulmonary interstitial fibrosis secondary to a large amount of inhaled foreign material. Analysis of a sample of the lung by X-ray diffraction identified the crystalline material as magnesium silicate hydroxide ($Mg_3Si_4O_{10}(OH)_2$), the mineral talc (Fig. 5).

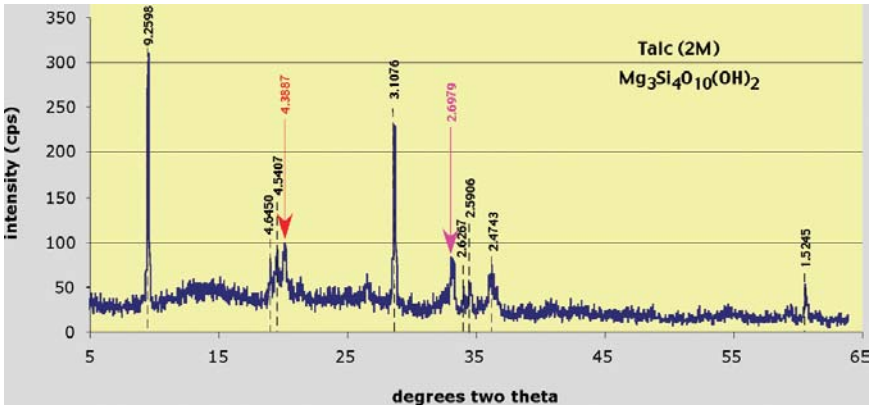

FIG. 5—*Bulk, unoriented, X-ray diffractogram of material recovered from lung tissue showing eight unit cell measurements (eight labeled peaks without arrows) definitive for talc. The two peaks labeled with arrows at 4.3887 and 2.6979 Angstroms are probable talc contaminants or oxidation products of sample processing.*

Discussion

Factitious disorders include those conditions in which an individual falsifies medical history and/or produces or simulates illness for no apparent purpose other than to assume the role of a sick person. Factitious disorders can be divided into three subtypes: factitious disorder with psychological symptoms, chronic factitious disorder with physical symptoms, and atypical factitious disorder (1). Although originally used in 1951 by Asher to describe patients with factitious illness who make migratory hospitalization a way of life, more recently Munchausen syndrome has come to be used as synonymous with chronic factitious disorder with physical symptoms (2,3). The prevalence of Munchausen syndrome is difficult to ascertain, due in part to the peripatetic nature of the patients (3). Munchausen syndrome patients are approximately equally distributed between males and females, and usually manifest pattern behaviors before age 20, but have been reported up to age 62 with a mean range of 35 to 39 years (3,4). A Munchausen syndrome "career" is thought to last around nine years, although some patients have continued for as many as 50. Persons with Munchausen syndrome frequently have training in medically-related professions (4) or are closely associated with someone who does, such as a family member (3). Their medical knowledge facilitates the production of plausible clinical histories, complete with details supportive of their "illness". Munchausen syndrome patients often consent to, and sometimes demand, invasive, often dangerous procedures (5). Therapy for Munchausen syndrome is difficult, with insight-oriented psychotherapy and behavioral modification resulting in some benefit in selected patients (6).

The presentation of Munchausen syndrome is limited only by the medical knowledge and imagination of the patient (7), and symptoms involving essentially every organ system have been reported, including the respiratory system (4). Although they are an unusual presentation for Munchausen syndrome, pulmonary manifestations may include hemoptysis, bronchospasm, chronically low oxygen saturation, pulmonary embolism, and subcutaneous emphysema (2,8). The symptoms and signs may be so convincing that thoracotomies and lung resections are often contemplated (9) and sometimes undertaken (10).

In contrast to hemoptysis and bronchospasm, cystic fibrosis is a decidedly uncommon respiratory presentation for Munchausen syndrome. This is thought to be due at least in part to the complicated nature of the disease; associated signs and symptoms can be difficult to feign, and the requisite laboratory studies are not easily manipulated (7). Cystic fibrosis is the most common potentially lethal inherited disorder in the Caucasian population and should be suspected in patients with a variety of findings, including failure to thrive, steatorrhea, recurrent respiratory infections, wheezing, obstructive lung disease and digital clubbing. The diagnosis can be confirmed by an elevated sweat chloride test result or by the identification of a mutation within the cystic fibrosis transmembrane regulator (CFTR) gene on chromosome 7 (5) .

Most case reports of factitious illness presenting as cystic fibrosis involve cases of Munchausen syndrome by proxy. Recognized in 1977, Munchausen syndrome by proxy involves a third party, usually a mother, providing spurious information about, producing physical findings in, and/or adulterating the laboratory samples obtained from the patient, usually a child (6,11). The first reported case of Munchausen syndrome *not* by proxy presenting as cystic fibrosis was described by Rusakow et al. in 1993 (5). In this case, a 19-year-old white female claiming to have cystic fibrosis presented to the emergency department with hemoptysis, dyspnea, fever, and a productive cough. The patient was evasive regarding details of her past medical history, and was reluctant to undergo a sweat chloride test or genetic evaluation. When she did consent to a sweat chloride test, she was not monitored continuously; levels from each arm were low and not confirmatory of cystic fibrosis, and there was a 37.4 mEq/L disparity between the left and right arms. The patient was diagnosed with Munchausen syndrome, and a review of medical records from other institutions revealed that Munchausen syndrome was suspected on several previous occasions.

In 2002, Highland and Flume (7) reported another case of Munchausen syndrome presenting as cystic fibrosis. A 25-year-old white woman with some medical training arrived at their adult cystic fibrosis program for further care. She reported that she was diagnosed with cystic fibrosis at the age of nine. Her medical records contained a technician's worksheet reporting an elevated sweat chloride level of 89 mEq/L; however, the final computerized report showed the value as 39 mEq/L. Closer inspection of the technician's worksheet showed that it clearly had been altered. Genotype analysis for 70 cystic fibrosis mutations was negative. When she was told she may not have cystic fibrosis, and that additional testing would be required, she moved out of state. Subsequently, her case was presented at a North American cystic fibrosis conference, where it was recognized that a patient with an identical history had presented at another cystic fibrosis center.

The present case provided a unique opportunity for clinical-pathologic correlation in a Munchausen syndrome patient whose

"career" spanned at least several years. Her history was typical of Munchausen syndrome patients in that she had medical training as a nurse, she was suspected by medical staff of sabotaging prescribed treatments, and she repeatedly refused laboratory testing to support her claims of cystic fibrosis. In addition, she had undergone several invasive procedures, including partial pneumonectomy. However, in contrast to the other Munchausen syndrome cases presenting as cystic fibrosis, this patient did have legitimate and marked respiratory compromise, but one that was factitious rather than genetic in origin. Her severe pulmonary interstitial fibrosis was the result of inhaling a large quantity of talc over an unknown period of time, a phenomenon that has been described in other Munchausen syndrome cases. Egan et al. (12). reported a case of a Munchausen syndrome patient with self-induced inhalational pulmonary talcosis originally diagnosed as asthma. In that case, an open lung biopsy showed birefringent golden crystals in the respiratory tree, some of which were engulfed by multinucleated giant cells. The patient later admitted to inhaling large amounts of baby powder.

Death from Munchausen syndrome is rare, and most instances of mortality from factitious illness are Munchausen syndrome by proxy cases (13). Nichols et al. (13) reported the case of a 31-year-old white female hospital security guard who, after presenting with chills, fever and headaches, entered a restroom, where she was later found dead. Among the items found on her person were a brown bag of off-white powder and a 10 mL syringe containing 3 mL of an off-white slurry; analysis proved these substances to be corn starch. A full autopsy was conducted, in which at least three recent left anticubital injection sites were discovered. Microscopy performed on the lung sections demonstrated multiple intraarterial and intracapillary partially refractile crystalline starch granules, displaying the typical corn starch "Maltese cross" under polarized light.

While the clinicians in the present case suspected that the patient's severe pulmonary disease was self-inflicted, the mechanism by which the damage was rendered remained a mystery during her life. The identification at autopsy of copious amount of birefringent foreign material in the patient's lungs was an unexpected and illuminating discovery. Not only did the limited autopsy confirm the clinical impression of severe chronic lung disease secondary to Munchausen syndrome, but the nature of the foreign material and the route of administration were also deduced. When injected, talc should be found within the pulmonary vasculature, as well as in perivascular and interstitial locations (14); the presence of talc within airspaces coupled with the absence of intravenous talc supports a respiratory route. One might suspect that the same foreign material should have been identified in the lung resection specimens, but no material pertaining to those surgical specimens was available for review, and no histologic findings other than "abscesses" were mentioned in the medical record.

Most reports of Munchausen syndrome patients appear in either internal medicine or emergency medicine literature, with only a paucity of information in the pathology literature. This case demonstrates the value of the autopsy, even a limited autopsy, in suspected cases of Munchausen syndrome. The diagnosis of Munchausen syndrome can be difficult, and whether in the laboratory or in the autopsy suite, pathologists should be aware of the vital role they can play in recognizing this sometimes fatal condition.

Acknowledgments

The authors would like to thank Michael Crossey, MD, PhD, for facilitating the analysis of the lung sample by X-ray diffraction. Powdered samples were analyzed by X-ray diffraction (XRD) in the XRD Laboratory in the Department of Earth and Planetary Sciences at the University of New Mexico, using a Scintag Pad V diffractometer with DataScan 3.1 software (from MDI, Inc.) for system automation and data collection. Cu-K-alpha radiation (40 kV, 35 mA) was used with a Bicron Scintillation detector (with a pyrolitic graphite curved crystal monochromator). Data were analyzed with Jade 5 Software (from MDI, Inc.) using the ICDD (International Center for Diffraction Data) PDF2 database (rev. 1999) for phase identification.

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