Ethical Considerations in REM Sleep Behavior Disorder

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ABSTRACT

A patient diagnosed with REM behavior sleep disorder (RBD) has as much as a 65% risk of developing an α-synucleinopathy. Currently, it is not possible to predict whether an individual will develop a disease, or, if so, which disease.The neurologist treating the patient must consider (1) the difference between disclosing a diagnosis and disclosing the risk of a diagnosis; (2) whether to disclose this risk to patients; and (3) if deciding to disclose the risk, the appropriate timing of such a conversation.

Continuum (Minneap Minn) 2013;19(1):199–203.

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Relationship Disclosure:
Dr Vertrees reports no disclosure. Dr Greenough has received a grant from Dartmouth Center for Clinical and Translational Science Pilot and Collaborative Studies Program.

Unlabeled Use of Products/Investigational Use Disclosure:
Drs Vertrees and Greenough report no disclosures.

Case

Note: This is a hypothetical case.

A 70-year-old man presented to the clinic at the insistence of his wife, who noted the gradual development of strange behaviors at night, including shouting, swearing, kicking, and punching. Her husband had been a sleeptalker and restless sleeper for many years. More recently, the episodes had become more violent, and his wife had bruises on her left arm and leg as a result of his nocturnal movements. She now slept in a different room.

The patient was somewhat embarrassed. He was aware of nightmares and reported dreaming of muggers attacking him and his wife. In the dream, he would fight the assailants. Much to his chagrin, when he would awaken he would find he had been punching his wife. He had also injured himself, recently falling out of bed and sustaining a laceration over his eyebrow.

Polysomnography revealed frequent periodic limb movements in non-REM sleep and elevated motor tone in REM sleep, which in conjunction with the history were consistent with the diagnosis of REM sleep behavior disorder (RBD). Because RBD is associated with an increased risk of developing a neurodegenerative disease, the neurologist felt that the risk should be disclosed to the patient and his wife, but a colleague advised that this would be unwarranted as it would only cause worry for them. This case raises the following ethical questions:

1. Should the physician disclose the risk of neurodegenerative disease in patients with RBD? If so, how? If not, why?
2. Does the disclosure of a diagnosis differ from the disclosure of a risk?

DISCUSSION

RBD is a parasomnia characterized by dream enactment behaviors during REM sleep, including excessive motor activity and vocalizations. RBD is known to be a common clinical feature in the α-synucleinopathies (including Parkinson disease [PD], dementia with Lewy bodies, and multiple system atrophy), and it can also be seen in spinocerebellar ataxia type 3, Huntington disease (HD), and other neurologic conditions. Several reports have shown that patients who are diagnosed with idiopathic RBD (iRBD) and who have no clinical signs or symptoms of the α-synucleinopathies are at a significantly increased risk of developing one of the α-synucleinopathies later in life. Schenck and colleagues first reported the development of PD in 38% of patients originally diagnosed with iRBD.[1] Later studies show the risk of developing neurodegenerative disease ranges from 45% to 65%, with higher rates seen the longer patients are followed.[2–4]

A significant delay from the onset of the RBD symptoms to the onset of the α-synucleinopathy exists. For men initially diagnosed with iRBD over the age of 50, the mean time to onset of the α-synucleinopathy is 13 years.[5] The link between neurodegenerative disease and RBD may be related to the pathologic involvement of common brainstem structures, including the nigrostriatal complex, locus coeruleus, raphe nucleus, and others.[6] Furthermore, approximately 50% of patients with RBD have mild cognitive impairment, and RBD is associated with cognitive decline in PD.[7]

The association between RBD and neurodegenerative disorders raises the issue of disclosure of potential risk for patients presenting with RBD who have no signs or symptoms of neurodegenerative disorders, an issue that has not been addressed in the peer-reviewed literature. The difficulty lies in the difference between disclosing a diagnosis versus disclosing the risk of a diagnosis. The diagnosis of RBD is not absolutely predictive of the development of a neurodegenerative disease, but rather suggests an increased susceptibility or probability (compared to the general population) that the patient will develop such a disorder in the future. Clinical examples with similar characteristics include genetic susceptibilities toward dementia and for sudden unexplained death in epilepsy (SUDEP).[8,9]

The ethical principles of autonomy, informed consent, and respect for persons support disclosure of information to patients.[10] In RBD, however, a patient's individual risk of developing a neurodegenerative disorder is uncertain, and physicians are unable to provide definitive information. By sharing the implications of the diagnosis of RBD with patients, physicians enable them to make an informed decision, thus preserving patients' autonomy. Because decisions are based on general rather than specific probabilities, both patient and physician may be frustrated, but the potential frustration from uncertainty is an inadequate justification for withholding information the patient needs to make an informed decision. Furthermore, withholding information may harm the doctor-patient relationship, which relies on veracity, the ethical principle, and the physician's duty to tell the truth.[10]

Does disclosing the risk to the patient benefit the management or prevention of the disorder for which the patient is at risk? With SUDEP, disclosure of the risk may help prevent sudden death by promoting compliance in taking antiepileptic medications.[9] With RBD and neurodegenerative diseases, however, some physicians may find it pointless to disclose the risk of developing disease

because no interventions exist to prevent or delay disease development, and no definitive disease-altering treatments exist. Thus, some physicians argue against disclosing information that would disturb a patient's current life unless and until signs of neurodegenerative disease develop. The patient might live many years awaiting the onset of a disorder that may never occur. By sharing the risk of developing an α-synucleinopathy, they would argue, the patient may experience years of needless worry or anxiety as they anticipate developing a neurodegenerative disease. Many physicians seem to share this view, as dementia diagnoses are withheld from patients in about 50% of cases.[11] This is in part done to shield patients from worry about developing a dementia.

Recent data support disclosure of probabilities of a diagnosis on the basis of the principle of beneficence. Up to 92% of patients with dementia and their family members desire to know diagnoses as soon as possible.[12,13] Reasons include putting affairs in order, such as wills and advance directives; arranging for assistance with housing, caregiving, and finances; initiating treatment as soon as possible; and indicating preferences for treatments and research participation. The aforementioned actions are best done while the patient retains decision-making capacity.[12–15] Similar benefits would be found with early disclosure of the risk of iRBD. The physician, patient, and family can discuss the implications of developing an α-synucleinopathy, and if desired the patient can seek a second opinion or referral to a subspecialist.

In addition, disclosure to patients and family members alerts them to watch for the onset of symptoms such as tremor, bradykinesia, memory impairment, or orthostasis, which may allow for early diagnosis. If neurodegenerative disease is diagnosed, therapies can be initiated for enhancing quality of life. If a definitive neuroprotective agent for α-synucleinopathies was identified, this is a population that might benefit from early treatment with this therapy. The knowledge that most patients have a desire to know what is wrong with them and that beneficial interventions exist if an α-synucleinopathy develops may sway physicians to disclose the risk of a neurodegenerative disorder. The physicians might otherwise be reluctant to disclose information, believing that patients have little to gain.[15]

Even if the physician chooses not to discuss the association of iRBD and α-synucleinopathies, the patient and family are likely to discover this information on the Internet (eg, *www.scientificamerican.com/blog/post.cfm?id=is-rem-sleep-disorder-early-sign-of-2008-12-24*), which would also likely lead them to conclude correctly that the physician had withheld important information from them, which violates the principles of truth telling and respect for autonomy. Patients may have grounds for legal action against physicians if information is withheld, unless there is overwhelming evidence that receiving such information is harmful, invoking the so-called therapeutic privilege.[10] Depending on the website, the patient may be exposed to misinformation.

Could disclosure of the future risk of neurodegenerative disease be harmful? The risks of stigmatization, developing hopelessness and despair, suicidality, or losing personal identity have been claimed as reasons not to disclose the diagnosis of Alzheimer dementia.[16] One model for disclosure of neurodegenerative processes has been with HD, which has been associated with RBD; however, the paradigm of disclosure is different from iRBD because the risk can be more accurately estimated based on HD's genetic nature and available

testing.[6,8] Disclosure of HD genetic status and disease risk has been known to cause anxiety, depression, and disruption of marriages and interpersonal relationships in presymptomatic HD patients as well as in undiagnosed individuals who have been notified of the risk of developing disease before genetic testing is performed.[17–19] Research in HD and dementia has shown that the harmful effects of disclosure are short-lived and that patients lacked signs of long-term psychological distress.[12,19]

Whether to break such news to patients should not be the question. Instead, the determination of when and how to do it is important.[8,20] Early disclosure appears to be the best approach rather than waiting until symptoms develop. But how early? This may depend on the individual patient. Disclosures should always occur in a patient-centered manner. The physician should determine what the patient knows about the disease and the risks it poses, and then build on the patient's knowledge and address any misconceptions. The conversation should focus on the goals of care. Emphasis should be placed on the availability of the physician for regular monitoring and, should disease develop, for symptomatic treatment and disease management.[14,21] Patients are likely to be reassured when physicians express their availability to care for them if an α-synucleinopathy were to develop. This approach, consistent with the principle of nonabandonment, is key. Neurologists should commit to closely follow patients to look for signs of neurodegenerative disease, allowing for early diagnosis and initiation of available therapies.

Finally, some patients may not want bad news shared with them. An appropriate approach would be to first ask patients if they want to know the future implications of their RBD diagnosis. If the patient expresses a desire to be informed, a discussion of neurodegenerative disease risk is relevant, and patients should be warned that bad news is coming. Patients may decline such a discussion when first offered. Patients have an autonomous right to refuse to hear this bad news.[22] If the patient initially declines such a discussion, the issue can be raised at a future visit once the patient has had time to digest the diagnosis of RBD. If the patient continues to decline, expressing one's availability to have such a discussion in the future when and if the patient so desires might be a reasonable next step.

RECOMMENDATIONS

Based on ethical principles and experience with other diseases, the patient in the case would benefit from the disclosure of the risk of developing neurodegenerative disease when informed of his RBD diagnosis. Telling the truth about the future risk of neurodegenerative disease with RBD while being honest about the uncertainty of the risk promotes his autonomy, is beneficent, and will engender the patient's trust in the physician. Furthermore, discussing the diagnosis of RBD and offering to discuss the potential long-term implications would be appropriate.

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