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The Spiral Axis as a Clinical Tool to Distinguish Essential Tremor from Dystonia Cases

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Abstract

Background: Tremor is a common feature of a variety of neurological disorders. In genetic studies of essential tremor (ET), investigators need to screen potential enrollees by mail or telephone to exclude those with other neurological conditions, especially dystonia. In clinical settings, the differentiation of ET and dystonia may also be very challenging. We hypothesized that the spiral axis, described below, is a useful screening tool to distinguish ET cases from dystonia cases.

Methods: We analyzed the hand-drawn spirals of 135 individuals enrolled in a genetics study at Columbia University Medical Center. Each of the four spirals was assessed for the presence of a single identifiable tremor orientation axis, and a spiral axis score (range = 0 – 4) [a single axis on all 4 spirals] was assigned to each enrollee.

Results: There were 120 ET cases and 15 cases with dystonic tremor. Most (101/120, 84.2%) ET cases had an axis score ≥1 vs. only half (8/15, 53.3%) of the dystonia cases (p =0.02). Receiver Operator Curve (ROC) analysis revealed that the use of a spiral axis score ≥2 as a cut off would exclude 60.0% of dystonia cases while including 67.5% of ET cases.

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Conclusion: Handwritten spirals appear to have a single predominant axis in more ET than dystonia cases. The evaluation of this axis has moderate diagnostic validity as a screening tool to distinguish ET cases from those with dystonia. Although this study did not assess the utility of this tool in clinical practice settings, future studies should do so.

Keywords

Essential tremor; dystonia; spiral; axis

Introduction

Tremor is a common feature of a variety of neurological disorders, including essential tremor (ET), Parkinson's disease (PD), and dystonia [1]. In genetic studies of ET, investigators must screen potential enrollees (i.e., reportedly affected relatives) by mail or telephone to ensure that they do not have other neurological conditions. Many individuals who self-report a diagnosis of "ET" actually have another neurological disorder rather than ET [2]. One previous study on misdiagnosis of tremor disorders reported that of the 26 patients with false ET, 6 (23.1%) were diagnosed with dystonia [2]. Another study of the over-diagnosis of ET demonstrated that 4 (40%) of 10 ET patients had dystonia [3].

To save time and resources, and avoid costly field trips to individuals who do not have ET, it is important to develop methods, aside from self-report, to effectively screen/exclude individuals with dystonic tremor prior to an in-person study visit. We previously reported [4] that on spiral drawing, the wave forms of ET cases generally aligned along a single predominant axis rather than several axes (Figure 1). To our knowledge, no study has investigated the spiral axis as a screening tool to distinguish ET from dystonia cases. Here, we analyzed the pre-enrollment hand-drawn spiral drawings of 135 individuals enrolled in a genetic study, all of whom self-reported ET. Each of the four spirals was assessed for the presence of a single identifiable tremor orientation axis, and a spiral axis score (range = 0 – 4) [a single axis on all 4 spirals] was assigned to each enrollee (Figure 1). After enrollment, each enrollee underwent a neurological examination, and 120 were diagnosed with ET and 15 with dystonic tremor. We hypothesized that the spiral axis would be a useful screening tool to distinguish ET cases from dystonia cases.

Methods

Participants

This study was conducted as part of the Family Study of Essential Tremor (FASET), Columbia University Medical Center (CUMC), New York [5]. Prior to enrollment, individuals with familial ET who were reported to have tremor were required to submit a set of four Archimedes spirals (two right, two left), drawn on a standard sheet of paper, without resting their wrist on the paper. These were analyzed and rated by a senior movement disorders neurologist (E.D.L) to determine the features and severity of tremor [5]. Individuals were selected for an in-person clinical evaluation if (1) one or more of the spirals had a rating ≥2, (2) they did not report a diagnosis of dystonia or PD, and (3) the tremor was not thought to be due to a medication or another medical condition. Upon enrollment, the

FASET coordinator obtained written informed consent, approved by the CUMC Institutional Review Board, from all participants.

Clinical Evaluation

Enrollees underwent an in-person evaluation consisting of a series of questionnaires to gather demographic and clinical information. Each enrollee was asked to draw four Archimedes spirals (two right, two left) on a standard sheet of paper, without resting their wrist on the paper.

In addition to the questionnaires, enrollees took part in a videotaped neurological examination, which included assessments of postural, kinetic, intention, and rest tremors as well as other movements [5]. E.D.L. analyzed all videotaped examinations, assigning a total tremor score, ranging from 0 – 36 [5]. The severity of kinetic tremor during the drawing of each of the four spirals were given a score of 0 – 4 (0 = no visible tremor, 1 = low amplitude tremor, 2 = moderate amplitude, 3 = large amplitude, 4 = extremely large tremor) [5], and the spiral severity score was the average of the four ratings (range = 0 – 4). During the examination, torticollis, spasmodic dysphonia, blepharospasm, and other dystonic postures or tremors were specifically assessed, as were signs of parkinsonism [5].

After a thorough assessment of both the videotaped neurological examination data and questionnaires, ET diagnoses were assigned by E.D.L. based on published diagnostic criteria [5]. A dystonia diagnosis was assigned when there were sustained muscle contractions, which caused abnormal postures, or twisting movements [5].

Spiral Axis Rating

Blinded to clinical information, E.D.L. analyzed the screening spirals of all 135 study enrollees that were screened between October 2011 and February 2013. Each of the four spirals was assessed for the presence of a single identifiable tremor orientation axis (rather than the presence of multiple visible axes or no clear axis), and a spiral axis score (range = 0 – 4) [a single axis on all 4 spirals] was assigned to each enrollee (Figure 1). In a secondary analysis, he also rated the second set of spirals (i.e., the in-person spirals).

Statistical Analyses

Statistical analyses were performed in SPSS (Version 21). Student's t-tests, chi-square (x^2) tests, and Fisher's exact tests were used to test for statistical significance. The spiral axis score was not normally distributed (Kolmogorov-Smirnov test, $z = 1.98$, $p < 0.001$); therefore, non-parametric tests (Mann-Whitney test, Spearman's rho) were used when assessing this variable. In one analysis, we stratified the sample by the spiral severity score. To assess the effects of potential confounders of the association between spiral axis score and diagnosis (ET vs. dystonia), adjusted logistic regression models were performed. Receiver Operator Curve (ROC) analysis was also performed to assess the diagnostic utility of the spiral axis score, examining a full range of cut-off points.

Results

There were 120 ET cases and 15 dystonia cases (6 with torticollis, 2 with arm dystonia, 1 with blepharospasm, and 6 with dystonia in more than one region). The dystonia cases had not reported a diagnosis of dystonia during the screening phase of the study – dystonia was diagnosed after enrollment based on the videotaped neurological examination. ET cases were on average 7.1 years younger than dystonia cases (p = 0.03, Table 1). The proportion of women was 50.8% in ET cases and 93.3% in dystonia (p = 0.002, Table 1). Otherwise, the two groups did not differ by race, duration of tremor, or presence of voice or cranial tremor on examination (Table 1).

The spiral axis score was higher in ET cases than dystonia cases (2.03 ± 1.31 [median = 2] vs. 1.20 ± 1.37 [median = 1], p = 0.03, Table 1). Spiral axis score was associated with age in years (Spearman's r = 0.19, p = 0.03), but not with age of tremor onset (Spearman's r = −0.04, p = 0.69). Spiral axis score did not differ by race (white vs. non-white, Mann Whitney test p value = 0.96). Spiral axis score was higher in men (2.32 ± 1.23, median = 2) than women (1.64 ± 1.35, median = 2), Mann Whitney test p value = 0.003. There was a correlation between spiral axis score and duration of tremor (Spearman's r = 0.27, p = 0.002) and total tremor score (Spearman's r = 0.47, p <0.001). There was no association between spiral axis score and presence of voice tremor on examination (Mann Whitney test p value = 0.29) or presence of cranial (neck, voice, or jaw) tremor on examination (Mann Whitney test p value = 0.41).

As noted above, there was an association between spiral axis score and tremor severity. To ensure that the ET vs. dystonia difference in spiral axis score was observable across all levels of tremor severity, we stratified ET and dystonia cases into tertiles based on the spiral severity score: Tertile 1 = spiral severity ≤ 2.5; Tertile 2 = spiral severity 3 – 3.5; Tertile 3 = spiral severity = 4.0. In each of the tertiles, the mean spiral axis score was numerically higher in ET cases than dystonia cases: 1.10 ± 1.09 (ET) vs. 0.43 ± 1.13 (dystonia) in Tertile 1; 1.96 ± 1.17 (ET) vs. 1.80 ± 1.48 (dystonia) in Tertile 2; and 2.78 ± 1.17 (ET) vs. 2.00 ± 1.00 (dystonia) in Tertile 3. Since the sample size in each tertile was small, we are not reporting p values.

To further assess the reliability of this screening tool, we also rated the second set of spirals (i.e., the in-person spirals). The correlation between the pre-screen spiral axis score and the in-person spiral axis score was high (Spearman's r = 0.88, p < 0.001).

In an unadjusted logistic regression analysis, spiral axis score was associated with diagnosis (ET vs. dystonia) (odds ratio [OR] = 1.67, 95% confidence interval [CI] = 1.06 – 2.63, p = 0.027). We considered potential confounders, identified in bivariate analyses, and performed logistic regression analyses adjusting for these variables. In the first model, we adjusted for age and gender. In our second model, we adjusted for a larger number of variables: age, gender, age of tremor onset, duration of tremor, and total tremor score. In the final adjusted logistic regression model, spiral axis score was independently associated with the diagnosis (OR = 1.79, 95% CI = 1.12 – 2.86, p = 0.015, i.e., ET cases had higher spiral axis scores than dystonia cases); in the same model, age was also marginally associated with diagnosis (OR = 0.97, 95% CI = 0.93 – 1.01, $p = 0.06$).

Most (101/120, 84.2%) ET cases had a spiral axis score ≥1 vs. only half (8/15, 53.3%) of the dystonia cases (Fisher's $p = 0.02$). ROC analysis revealed that using a spiral axis score ≥2 as a cut off would exclude 60.0% of dystonia cases while still including 67.5% of ET cases.

Discussion

Despite attempts to use questionnaires to screen out enrollees with dystonia, in the current study, we still enrolled 15 dystonia cases, so there is a clear need for better screening methods. This need prompted these analyses. In an earlier study, we examined hand drawn spirals in a small sample of 29 ET cases and identified an axis in one or both spirals in 23/29 cases [4]. We now explore whether a tremor axis is identifiable in dystonia patients. Our current results show that an analysis of handwritten spiral drawings is a moderately useful screening tool to distinguish ET cases from dystonia cases during the pre-enrollment phase of a genetic study, and it is likely to be useful in other studies (e.g., epidemiological studies, clinical studies). This screening tool is not meant to replace in-person evaluations, but rather, to serve as a screening method to save time and resources when conducting large-scale genetic studies.

Although the current method is certainly not perfect, ROC analysis revealed that using a spiral axis score ≥2 as a cut off would exclude 60.0% of dystonia cases while still keeping 67.5% of ET cases. This would help with achieving the goal of enrolling individuals with a cleaner subset of ET (i.e., ET without dystonia) in genetic studies. Hence, the method provides one more tool available to researchers.

Aside from its use in research studies, the current method may have some utility in clinical practice settings, although this was not directly assessed. Studies show that in clinical practice settings 30 – 50% of ET cases are mis-diagnosed [2,3]. These studies reported dystonia to be among the more common diagnoses among patients classified as having ET [2, 3]. The presence of a single predominant axis would be one clinical feature that would make the ET diagnosis more likely.

The physiological reason why ET cases are more prone to exhibit a single spiral axis and dystonia patients are not is not clear, however, dystonic tremor may have an irregular quality, and this could contribute to the phenomenon we observe. Studies have shown that a distinctive characteristic of tremor in patients with dystonia is the irregular and broader range of tremor frequency [6]. The extent to which this is responsible for our findings is not clear.

The study had limitations. First, we analyzed spirals drawn on paper. Use of a computerized spiral analysis may prove to be more precise, although is not practical for screening patients from afar. Second, all ET cases came from a single genetic study; additional studies should assess the spirals in the setting of non-familial ET.

In summary, handwritten spirals appear to have a single predominant axis in more ET than dystonia cases. The evaluation of this axis has moderate diagnostic validity as a screening tool to distinguish ET cases from those with dystonia. Although this study did not directly assess the utility of this tool in clinical practice settings, future studies should do so.

Acknowledgments

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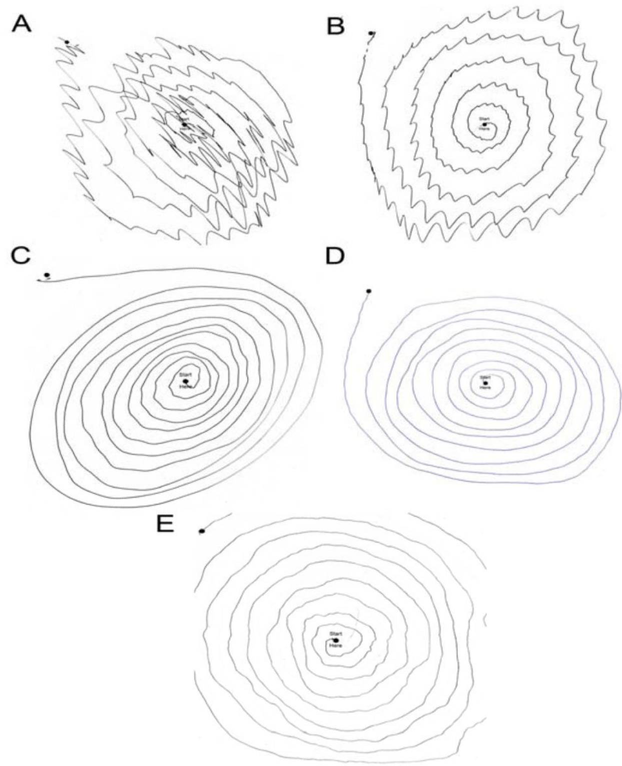

Figure 1.

(A-E) Examples of spiral drawings. As previously reported in ET, spirals drawn with the right hand typically have a single identifiable tremor orientation axis (i.e., tremor is most severe at 1 - 2 o'clock and 7 - 8 o'clock), whereas those drawn with the left hand have a single identifiable tremor orientation axis that is 90 degrees to that on the right (i.e., tremor is most severe at 10 - 11 o'clock and 4 - 5 o'clock)[3]. (A) Left hand spiral drawing of an ET patient with severe tremor. The predominant axis is oriented at 10 - 11 o'clock and 4 - 5 o'clock. (B) Right hand spiral drawing of an ET patient with moderate tremor. The predominant axis is oriented at 1 - 2 o'clock and 7 - 8 o'clock. (C) Right hand spiral drawing of an ET patient with mild to moderate tremor. The predominant axis is oriented at 1 - 2 o'clock and 7 - 8 o'clock. (D) Left hand spiral drawing of an ET patient with mild tremor. The predominant axis is oriented at 9 - 11 o'clock and 3 - 4 o'clock. (E) Spiral drawing of a patient with dystonic tremor; tremor is present but no single predominant axis stands out.

Table 1

Demographic and Clinical Characteristics of 120 ET cases vs. 15 Dystonia Cases

Characteristic	ET Cases (N = 120)	Dystonia Cases (N = 15)	Significance
Age (years)	58.4 ± 17.5	65.5 ± 10	t = 2.31, p = 0.03
Female gender	61 (50.8)	14 (93.3)	X^2 = 9.75, p = 0.002
White race	108 (90.0)	12 (80.0)	X^2 = 1.35, p = 0.38
Age of tremor onset (years)	26.9 ± 18.8 [median = 20]	37.5 ± 21.4 [median = 40]	Mann-Whitney = 1.83, p = 0.07
Duration of tremor (years)	31.6 ± 19.3 [median = 31]	26.5 ± 17.5 [median = 24]	Mann-Whitney = 0.75, p = 0.45
Voice tremor on examination	17 (14.3)	2 (14.3)	X^2 = 0.00, p = 1.0
Cranial tremor on examination	55 (45.8)	4 (26.7)	X^2 = 1.99, p = 0.18
Total Tremor Score	18.9 ± 6	16.8 ± 6	t = 1.23, p = 0.22
Spiral Axis Score	2.03 ± 1.31 [median = 2]	1.20 ± 1.37 [median = 1]	Mann-Whitney = 2.23, p = 0.03
Spiral Axis Score 0 1 2 3 4	19 (15.8) 20 (16.7) 42 (35.0) 16 (13.3) 23 (19.2)	7 (46.7) 2 (13.3) 3 (20.0) 2 (13.3) 1 (6.7)	X^2 = 8.75, p = 0.068

Values are means ± standard deviations [medians] or numbers (percentages).