THE INTELLIGENCE OF PATIENTS WITH FRIEDREICH'S ATAXIA

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Most references to the intellectual status of patients with Friedrich's ataxia comprise vague clinical impressions. General statements about this aspect of the patients were, however, made early in the history of the disease. Friedrich himself (1863) noted the absence of intellectual defect in his cases. This view held sway for a long time. Raymond (1898) emphasized "pas de troubles intellectuels" as a characteristic of this disorder, and Whyte (1898) went so far as to say, "Mental defect seems a much more important sign than the retention of the knee-jerk in making one doubt a diagnosis of Friedrich's ataxia." In much the same strain Ladame (1890) wrote, "Some authors think they (i.e., patients with Friedrich's ataxia) always suffer from an arrest of intelligence. The case which we relate proves the contrary."

Turning to individual case histories we find that cases with "average" or "good" intelligence have been described by many authors (Gowers, 1881; Taylor, 1894; Mackenzie, 1894; Clarke, 1889; Small, 1895; Speer, 1921).

Cases with notably superior intelligence have been described by several authors. Thus Tresider's (1893) case was head boy of a public school, and took an honours degree at Cambridge. Bramwell (1897) described his patient as being "very intelligent," and Moody (1910) described one of his patients as "very intelligent."

Low intelligence in patients affected with Friedrich's ataxia has been noted by Power (1882), Ogilvie (1908), Speer (1921), Walter and Roesse (1926), Schröder (1937). The "three cases of genotoxic idiocy associated with Friedrich's ataxia," described by Nolan (1895) and often quoted in this connexion, would appear, in the light of further investigations by Batten and Wilkinson (1913-14), to have been cases of Pelizaeus-Merzbacher disease.

Hanhart (1923) said that one encounters in these cases of Friedrich's ataxia a deterioration of intellect and affect. He saw no cases of idiocy, and severe grades of mental deficiency only where the disease had been of very early onset. He warned against easy guesses at the intelligence level of these patients. He said that, just as many

imbeciles are over-estimated intellectually by some deceptive verbal facility, so may the hereditary ataxias be underestimated by their reticence.

Bell and Carmichael (1939) wrote on this subject

"There is no doubt that patients . . . may retain their mental faculties unimpaired, and may occasionally show a mental alertness above the average; but a very considerable proportion of cases . . . do show signs of mental deterioration . . . when such defect occurs it is not a late symptom of the disease but is often conspicuous from the early stages of the affection and is not, in marked cases, demonstrably contributed to by the cultural limitations imposed on the patient by reason of the ataxic disease itself . . . A few cases, some half dozen perhaps, undoubtedly showed signs of rather severe mental deterioration, which was probably associated with their ataxia disease and might presumably be attributed to an extension of the pathological processes; . . . I considered the possibility of measuring the intelligence quotient on a series of patients but decided that tests made under state of nervous tension owing to the circumstances of our visit, and tests which were likely to be influenced by the cultural restrictions necessitated by the disease, could contribute no positive information concerning the question of mental deterioration in cases of hereditary ataxia, and might even be misleading."

The first attempt to apply standard tests to such cases seems to have been that of Landsbergen (1912) in three cases of familial cerebellar ataxia. These tests were for memory, conceptual ability, and comprehension. There was no scoring given, tests and answers being quoted verbatim. The patients were of very low intelligence. Since then only two other investigations along quantitative lines seem to have been made, that by Sjögren (1943) and that by Blöchlinger (1946). The latter may be discussed first.

Blöchlinger was concerned to find, by tests of mental function, further evidence in support of Bleuler and Walder's (1946) view of the nature of mental change consequent upon diffuse brain damage at an early age. His material consisted of six chronic cases of Friedrich's ataxia which had been admitted to a general hospital purely on account of their physical disability. It is of interest to note

that one of these patients showed a frankly psychotic picture, with irritability, mood changes, and delusions of influence and persecution. Although these patients were given batteries of test questions, the answers to which are reported verbatim, no attempt is made to score these answers. Just as no measure of intellectual level which could be used for comparative purposes emerges, so there is no attempt to distinguish between present ability and a level of native intelligence from which the patient may have deteriorated during the course of his illness. Blöchlinger's conclusions concern chiefly conceptual ability, memory, and attention (said to be unchanged) and a slowing of thought, poverty of ideas, and tendency to perseveration. Critical ability, judgment, and native intelligence are said to be deteriorated.

Sjögren (1943) studied a long series of 188 cases of heredo-cerebellar ataxia; eighty-four of these being cases of Friedreich's ataxia. These were examined chiefly from the neurological and genetic standpoints, but some tests of mental function were given to these patients. "The extent of the testing is not too clear from Sjögren's account. One series of nine questions was put, of the type used in the comprehension section of the Wechsler scale. In addition some tests of serial subtraction were given, and as, in some cases only, he gives a mental age for the patient, one presumes that some other tests (not specified in the text) may have been given. There is no evident scoring, however, except for those cases where a figure is given for mental age. From his series Sjögren concluded that in Friedreich's ataxia: (1) 15 per cent. of cases showed oligophrenia; (2) 58 per cent. showed progressive dementia.

Most recently Knoepfel and Macken (1947) claim that ten out of fifteen cases showed feeble-mindedness, though they give no details whatsoever in all but two of these patients.

Clearly there has been much conflict of opinion in a region where clinical observation may be helpfully supported by more objective measurement.

Material and Methods

An investigation into the mental state of twenty cases of Friedreich's ataxia was carried out. Personality changes and frank psychoses occurred among these patients, and will form the subject of a later publication. In this present paper consideration is given to the intellectual status of these patients as gauged from their performance on various psychological tests, and from clinical estimates based on their behaviour and personal history (in particular their scholastic and work records).

Three of these patients were originally seen in the Maudsley Hospital, to which they had been admitted because of their mental state. The remaining seventeen

were either patients in London County Council hospitals for chronic sick (seven cases), confined to their homes (four cases), or at work (six cases). No selection was exercised in the choice of these patients; all who could be traced were investigated and recorded. In every case the diagnosis had originally been made by a neurologist of some distinction, and frequently the patient had been seen at various times by more than one such consultant. Each case was examined physically and mentally, relatives were interviewed, and in some cases a social worker visited employers to gather additional information.

Three psychological tests were used as a routine, though for various reasons it was not possible to apply all three to all the patients. The Mill Hill vocabulary test, Raven's progressive matrices, and the verbal half of the Wechsler Bellevue scale were the tests used. The Mill Hill vocabulary test (Raven and Walshaw, 1944) offers to the subject six alternative choice synonyms for each of the thirty-four test words. The test was originally standardized on school children and military personnel, and the results may be expressed in I.Q. Raven's progressive matrices test is very well known by reason of its adoption by the War Office for the testing of men in the General Service Corps in the 1939-45 war. It is a non-verbal test, highly saturated with "g"—the factor of general intelligence—and in consequence it tests present fluid ability with little reference to previous experience. The Wechsler-Bellevue scale comprises, in its verbal half, five sections (comprehension, arithmetic, digit repetition, similarities, and vocabulary), so that it tests intelligence from many angles.

These tests were regarded as complementary to clinical examination of the patient. By taking into account all the clinical data, including information as to school and employment record in each case, a clinical estimate was formed of intellectual status and the presence or absence of dementia. The matrix I.Q. expressed as a percentage of the Mill Hill I.Q. gave a conceptual quotient (C.Q.). C.Q.'s were obtained on eighteen patients who did both the Mill Hill and matrix tests. The Mill Hill test was unfortunately omitted in Case 2 (who subsequently died), whilst Case 17 was unable to read and, therefore, was unable to do the test at all. The Wechsler-Bellevue scale was applied in eighteen cases, being omitted for various incidental reasons in two cases.

The Mill Hill and matrix tests were also given to a miscellaneous group of fifteen chronic invalids, who had been in hospital for many years. Such invalids were afflicted with chronic rheumatism, heart disease etc., only those with diseases of the central nervous system being omitted. These patients were found in the wards of the hospital where were our Cases 7, 8, 9, and 10. The average age of the control group was higher than that of the Friedreich's ataxia group, and the average duration of invalidism was longer. Since the matrix test results were corrected for age the former factor was taken into account, and the longer duration of invalidism was a difference in the right direction, since the aim of using such a control group was to find how far any significant results of our testing could be explained simply as the result of invalidism, or could be related to

the particular disease (Friedreich's ataxia) affecting the group in which we are interested.

Patients in hospitals were visited there and tested; patients who were able to do so came up to the out-patients department for examination, those confined to their homes were visited for this purpose.

The choice of these particular tests was determined by several factors. Time was important, since many of these patients were visited in their homes and visits could not be prolonged to the point of interfering with domestic routine. Physical incapacity of the patients was another important factor. On this account Koh's blocks could not be used as a test, since some of the patients lacked the necessary power to lift and arrange the material. On the whole the tests selected worked well in these respects, and all the patients except one were able to use a pencil well enough to write a number or underline a word.

One final point need be mentioned here. All tests were untimed, so that the slowness in speech and response so well known in these cases was not in itself a factor influencing a test-score adversely.

Results

The results of intelligence testing are given in Tables I, II, and III. No patient showed idiocy or imbecility, nor did any patient show a sufficient degree of mental defect in the statutory sense to justify the use of that term in describing them.

Reference to Table I shows that only two cases (Nos. 6 and 17) had an I.Q. of less than 75 on the Mill Hill test. One of these (Case 17) was unable to read and, therefore, had no score on the test, but even including him the figure is not significantly different from the approximate 5 per cent. of the general population who would fall below I.Q. 75 on this test. On the Wechsler Test (a slightly different group) no patient had an I.Q. level of less than 75. From these observations it is possible to conclude that there is no evidence that mental deficiency is commoner among cases of Friedreich's ataxia than among the general population.

This point may be put to the test in another way, though the smallness of our group is not very conducive to the provision of strong proof. The mean I.Q. of the series, on the Mill Hill test, is 101.2, with a standard deviation of 13.79 and a standard error of 4.12. This is not significantly different from the population mean of 100. Here again there is no evidence to support the view that the initial or native intelligence of people who develop Friedreich's ataxia is significantly different from that of the general population.

In considering the more difficult question of the occurrence of dementia, reference must first be made to our clinical reports. Dementia presents itself to clinical observation in loss of recent memory, intellectual weakness (the inability to make the fine

TABLE I
FRIEDREICH'S ATAXIA: 20 CASES

Case no.	Sex	Age (years)	Age at onset (years)	Duration (years)	Mill Hill I.Q.	Matrix I.Q.	C.Q.	Wechsler I.Q.
1	M	15	13	2	97	90	93	104
2	F	21	19	2	—	66	—	75
3	F	23	11	12	100	83	83	—
4	F	24	14	10	114	95	83	96
5	M	27	14	13	119	98	82	—
6	M	35	21	14	71	73	103	78
7	F	39	13	26	122	118	97	115
8	F	38	20	18	89	74	83	81
9	F	43	21	22	97	95	100	101
10	F	35	19	16	115	97	84	111
11	M	25	11	14	100	94	94	108
12	M	45	14	31	92	70	76	95
13	F	27	13	14	92	88	96	94
14	F	32	13	19	122	117	96	117
15	M	28	17	11	108	104	96	114
16	M	28	13	15	108	95	88	110
17	M	18	10	8	—	71	—	—
18	M	29	10	19	86	69	80	—
19	M	23	10	13	97	88	91	—
20	F	26	12	14	92	80	87	—

comparisons and discriminations of the healthy mind), and disturbances of mood, such as euphoria. Later the loss of acquired good habits in relation to feeding and dressing may be obvious. In no case was any gross dementia clinically appreciable, but minor indications of such a process were evident in some of the patients examined. Thus Case 17 showed some impairment of recent memory as compared with old, and loss of attention and concentration in the serial 7 test. Some euphoria was observed. Conceptual quotient was 83. Case 10 showed some impairment of attention and concentration on the serial 7 test, and marked euphoria. Conceptual quotient in her case was 84. Case 12 showed poor ability to recall a name and address after five minutes, and poor general knowledge in face of a relatively high score on the Mill Hill test (I.Q. of 92). The conceptual quotient here was 76.

In certain other cases (182) it was suspected that dementia was present, but it was not possible to make this diagnosis clinically for various reasons (thus Case 18 was almost completely paralysed, whilst Case 2 had long periods of confusion while in hospital).

Sufficient has been said here, however, to enable us to draw the conclusion from clinical examination that some dementia—of no great degree for the most part—occurred in some three or four of our series, and was suspected in others.

If now we turn to Table I, it will be observed that the C.Q. is below 100 in all but two cases. Considering only those eighteen cases who did both Mill Hill and matrix tests the mean C.Q. is 90, the standard deviation is 8.1, and the standard error 1.9. The first point of interest here is the low standard deviation, which stamps the Friedreich ataxia patients as a remarkably homogeneous group in this respect. Secondly, the mean of 90 is significantly different from that of the general population (100), since Critical ratio = $\frac{100 - 90}{1.9} = 5.3$, a high degree of significance.

Similar comparison of the control group of general invalids (Table III) shows that the mean is within one of 100 so that there is no significant difference.

It would, therefore, seem that the deficit shown by the Friedreich group is not to be accounted for on the score of general invalidism. Clearly the low C.Q. in the cases of Friedreich's ataxia is due not to high Mill Hill scores, but to low scores on the matrix test. The I.Q.'s derived on these two

TABLE III
CONTROL GROUP OF 15 CHRONIC INVALIDS

Case	Age (years)	Duration (years)	Mill Hill I.Q.	Matrix I.Q.	C.Q.
D.W. ..	51	9	94	140	150
J.D. ..	40	12	115	101	88
G.F. ..	24	11	94	106	113
P.L. ..	20	7	97	110	114
E.G. ..	34	8	106	102	97
P.R. ..	24	24	92	95	103
E.A. ..	47	36	84	67	80
E.C. ..	57	12	100	114	114
M.B. ..	45	34	124	75	60
R.W. ..	47	12	97	106	109
C.A. ..	50	14	103	112	109
M.H. ..	51	1	81	92	114
N.P. ..	44	9	103	70	68
L.S. ..	39	10	74	75	101
F.A. ..	38	14	84	71	88
Means ..		14	96.3	95.7	100.5
S.D. ..		9.4	13.3	17.6	21.9

tests may be compared, since these two particular tests have the same standard deviation. The relevant figures for these I.Q.'s in all the Friedreich ataxia cases who did both tests is given here:

	Mean	S.D.	S.E.	Mill Hill	Matrix
	..	..	..	101.2	90.4
	..	..	..	13.79	12.34
	..	..	..	4.12	3.00

$$\text{Critical Ratio} = 3.3 \quad p = .002$$

This is additional support for the conclusion that the deficit as manifested in low C.Q. is not due to chance factors. Similar treatment of the Mill Hill and matrix I.Q.'s of the control group of general invalids shows no significant difference. There is no significant correlation between the extent of this deficit and the duration of illness or age of the patient. This would argue against the progressive nature of deterioration in these cases, and would accord with the clinical impression of Bell and Carmichael (already quoted) that where dementia occurs it tends to appear early in the disease.

The deficit so revealed might be explained on the basis of a specific defect. Cases of Friedreich's ataxia show, in addition to ataxia of the limbs, a frequent nystagmus. A specific disability for handling material in the form of geometrical patterns (as involved in the matrix test) cannot be ruled out of court in the absence of further evidence.

TABLE II

FRIEDREICH'S ATAXIA: 17 CASES
WECHSLER-BELLEVUE SCALE: WEIGHTED SUB-TEST SCORES

Case no.	Comprehension	Arithmetic	Digits	Similarities	Vocabulary	Total
1	12	6	7	11	13	49
4	8	9	6	9	12	44
6	6	6	3	5	6	26
7	12	12	9	12	14	59
8	10	3	4	4	8	29
9	9	9	9	8	10	45
10	14	10	6	14	11	55
11	14	9	6	14	11	54
12	8	9	7	7	8	39
13	7	10	6	11	8	42
14	13	13	7	14	14	61
15	11	17	9	12	11	59
16	14	10	6	14	12	56
17	8	9	6	5	5	33
18	8	6	4	5	6	29
19	11	9	10	9	9	48
20	9	6	11	8	11	45

to the contrary. Material derived from our Wechsler Bellevue tests (Table II), however, renders this view untenable. Here the five sub-test weighted scores have been given in detail for the seventeen patients who did this test. The means of the scores on digit repetition on the one hand, and vocabulary on the other, have been compared, and a critical ratio of 4.96 has been obtained. This is very highly significant.

It is generally accepted (Wechsler, 1944) that in testing patients who are demented this type of score, that is, a low digit repetition as compared with vocabulary score, is obtained. The difference is clearly not due to a high vocabulary score (see Mill Hill test results), so that we are justified in concluding a significantly low digit repetition score.

Weighing all the evidence of our psychological testing, we may conclude that a significant deficit is shown by the group of patients with Friedreich's ataxia, and such deficit is not to be accounted for merely by invalidism and the deprivation or lack of social opportunity which invalidism carries in its train.

Combining these test results with our clinical observations, which suggested dementia in a certain number of cases, we may justifiably equate such deficit with the occurrence of dementia.

We would, therefore, amend Sjögren's conclusions, which were by no means satisfactory in their derivation, by declaring that in our series of cases of Friedreich's ataxia there is no evidence to suggest that those who are marked down for the disease are intellectually different from the general population, but that patients affected with this disease show, as a group, significant dementia of no great degree.

Summary

The intelligence of twenty patients suffering from Friedreich's ataxia was measured by means of the matrix, Mill Hill, and Wechsler-Bellevue tests. A clinical estimate of intellectual deterioration was made in each case.

Similar testing was also carried out on a control group of seventeen chronic invalids from other causes.

No evidence was found to suggest that those marked down for Friedreich's ataxia are intellectually different from the general population, but patients affected with this disease show, as a group, significant dementia of no great degree.

This dementia appears early and is not progressive.

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