Quebec Cooperative Study
of Friedreich's Ataxia

Electroencephalographic Findings in Friedreich's Ataxia and Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)

R. W. BOUCHARD, J. P. BOUCHARD, R. BOUCHARD AND A. BARBEAU

SUMMARY: *Electroencephalographic studies have been done in two groups of hereditary ataxia: a group bearing the classical features of Friedreich's ataxia and a group clinically different described as autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). The qualitative anomalies observed in the two groups were similar and were comparable with the data reported in the literature. However, the main difference between the two groups is the greater incidence of EEG abnormalities in the ARSACS group, which suggests more involvement of the cortical and subcortical structures. This is reinforced by the lower I.Q. performance in the latter patients. Some comments are made about focal EEG findings, behavior and I.Q. In general, EEG was not considered a valuable instrument for diagnosis since no qualitative electric pattern could be identified. With regard to prognosis, EEG cannot be used as a criterion, since there is no relation between the degree of anomalies and the severity of the disease and since EEG does not worsen with the progression of the disease.*

RÉSUMÉ: *Nous avons étudiée le profil électroencéphalographique chez deux groupes d'ataxie héréditaire. Un groupe ayant les caractéristiques cliniques classiques de l'ataxie de Friedreich et un autre, différent sur le plan clinique, que nous avons récemment décrit sous le nom d'ataxie spastique familiale de Charlevoix-Saguenay. Les anomalies électroencéphalographiques relevées dans les deux groupes étaient qualitativement comparables et conformes aux récentes données de la littérature. Cependant nous avons trouvé une incidence beaucoup plus élevée d'anomalies chez les patients du groupe d'ataxie spastique familiale. Ceci suggère une atteinte plus marquée des structures corticales et sous-corticales chez ces patients et les moins bonnes performances aux épreuves psychologiques vont dans le même sens.*

Nous soulignons aussi quelques anomalies focales de l'électroencéphalogramme et leur signification possible, par rapport à certains aspects du comportement et aux résultats psychométriques chez nos patients. L'électroencéphalogramme ne s'est pas révélé très utile pour le diagnostic étant donné que nous n'avons pas défini de "pattern qualitatif" et également sur le plan pronostic, l'électroencéphalogramme n'apparait pas un examen très valable à cause du peu d'évolution de l'electroencéphalogramme avec l'âge, la progression et la sévérité de la maladie.

From l'Hôpital de l'Enfant-Jésus, Quebec City, and the Clinical Research Institute of Montreal.

Reprint requests for the complete supplement on Friedreich's Ataxia (Phase Two, Part Two) to:

Dr. André Barbeau, Clinical Research Institute of Montreal, 110 Pine Avenue West, Montreal, Quebec, Canada, H2W 1R7.

INTRODUCTION

Electroencephalographic (EEG) studies in hereditary ataxias have received little attention over the past decades. The initial report of Guillain et al. (1942) mentioned low amplitude cortical activity, frequent spikes, slow waves, and poor response to intermittent light stimulation (ILS) in six patients. In a study of fifteen patients with psychiatric symptoms associated with Friedreich's ataxia, Davies (1949 a,b) found six abnormal tracings, five of which showing temporal slowing. All of his patients had some intelligence testing and were found to be in the average range but, except for two well documented cases (Cases I and II), we do not know which of the patients had EEG abnormalities related to behavior and I.Q. performance.

In 1955, Kissel et al. reported epileptic activity in EEG's of three members of a "Friedreich family". It consisted of the mother and her two daughters. The youngest daughter, age 6, was unaffected. None of them had epilepsy, clinically. Badiu and Popescu-Tismana (1968) described EEG features of spino-cerebellar degenerations (Friedreich's ataxia, Roussy-Lévy's syndrome, and Pierre-Marie's ataxia) in fifteen patients and considered all their tracings abnormal, but without specific characteristic. Recently, Rémillard et al. (1976) and Andermann et al. (1976) reviewed EEG findings in a larger series of Friedreich's ataxia. These authors found 33% and 27.8%, respectively, of abnormal EEG's showing non specific mild anomalies, slow or irregular background activity, paroxysmal

rhythms, and poor driving response to ILS.

Our study concerns two groups of hereditary ataxic patients. Group I consists of patients with classical Friedreich's ataxia, all from the Rimouski area in Québec. Group II, clinically different, has been described and identified as ARSACS (Bouchard et al. 1978). Our aim has been to find out whether the Friedreich's ataxia patients had EEG profiles comparable to the groups previously studied and whether the ARSACS patients had different EEG findings, as the clinical picture might suggest. We also searched for correlations between the age of onset and the duration of the disease as well as the I.Q. performance. It has been demonstrated (Bouchard et al. 1978) that the ARSACS patients show a lower I.Q. performance than normal in many sub-tests of the non-verbal scale.

SUBJECTS, MATERIAL AND METHODS

EEG tracings were recorded in ten Friedreich's ataxia patients (Group I), four males and six females, and in nineteen ARSACS patients (Group II), seven males and twelve females. Seven patients of Group II had a second tracing done after a few months interval.

All the tracings were recorded in the same EEG laboratory with the International 10-20 System, using 8 or 16 channel Grass Model 6 electroencephalographs. Standard recording included bipolar and monopolar montages, hyperventilation, and photic stimulation. No other activation technique was used nor was any drug administered.

For Group I, the clinical features compatible with the diagnosis of Friedreich's ataxia are described elsewhere in this issue, as well as the psychometric evaluation. The age ranged from 17 to 28 with a mean of 22. All patients were right handed. For Group II, the clinical description and psychometric testing have been reported by Bouchard et al., (1978). All patients but three were right handed. The age ranged from 17 to 43 with a mean of 30. One female patient was thought to have epilepsy in her youth, but no clear history of seizure had been documented.

EEG tracings with well modulated symmetric alpha rythm, good alpha blocking to eye opening, and without diffuse or focal slowing or paroxysmal activity were considered normal. Minimal non focal dysryhthmias or occasional low voltage slowing over the temporal regions were not considered abnormal, nor were tracings showing only poor or no response to photic stimulation.

RESULTS

The results are summarized in Table I. In Group I, three patients (30%) had abnormal EEG, showing intermittent diffuse slowing or bursts of medium to high voltage slow waves (Fig. 1). One right handed patient, without obvious clinical lateralising signs, had focal slowing over the right temporo-parietal region. Six tracings (60%) showed poor unsustained occipital driving response to ILS. The other four (40%) had normal responses.

In Group II, twelve patients (63%) had abnormal EEGs. Of these, eleven showed background irregularities or intermittent slow activity. In three cases this was slightly more marked over the right hemisphere (right handed patients) and in one case over the left temporal region (left handed patient). Ten patients showed bursts of slow waves of subcortical origin, with paroxysms similar to the tracing shown in Figure 1 in four cases. No epileptic activity was recorded in the questionnably epileptic patient. Thirteen patients (68%) with either normal or abnormal EEG had poor driving response to ILS. The second EEG recorded in seven patients did not show any significant difference with regard to the usual parameters except for driving response to ILS. The occipital response was either much better or worse in five patients and in two of them, the response was strikingly opposite to the first tracing, i.e. a normal response at the first recording and a very poor at the second for one patient, and vice versa for the other patient.

No correlation could be drawn within each group between the incidence or the degree of EEG abnormality and the age, the sex, the duration, and the severity of the disease. EEG's of the youngest of either group compared with those of the oldest were similar, suggesting that the abnormalities either develop early in life or at the beginning of the disease and thereafter show little or no change or do not appear at all.

With regard to I.Q. and EEG (I.Q. details of Group I in this issue and Group II in our earlier paper), there was no correlation between EEG findings and the ability to handle verbal material. The verbal I.Q. was close to normal in both groups (Group I: 100.80; Group II: 92.67), although slightly lower in ARSACS. For non-verbal material, both groups had a

TABLE I

EEG Profile of Ataxic Patients

		Abnormalities			Overall Interpretation	
	Number of patients	Irregular background or slowing	Subcortical bursts or paroxysms	Poor driving to photic stimulation	Abnormal	Normal or within normal limits
Friedreich's Ataxia (Group I)	10	2	1	6 (60%)	3 (30%)	7 (70%)
ARSACS (Group II)	19	11	10	13 (68%)	12 (63%)	7 (37%)

https://doi.org/10.1017/S0317167100119626 Published online by Cambridge University Press

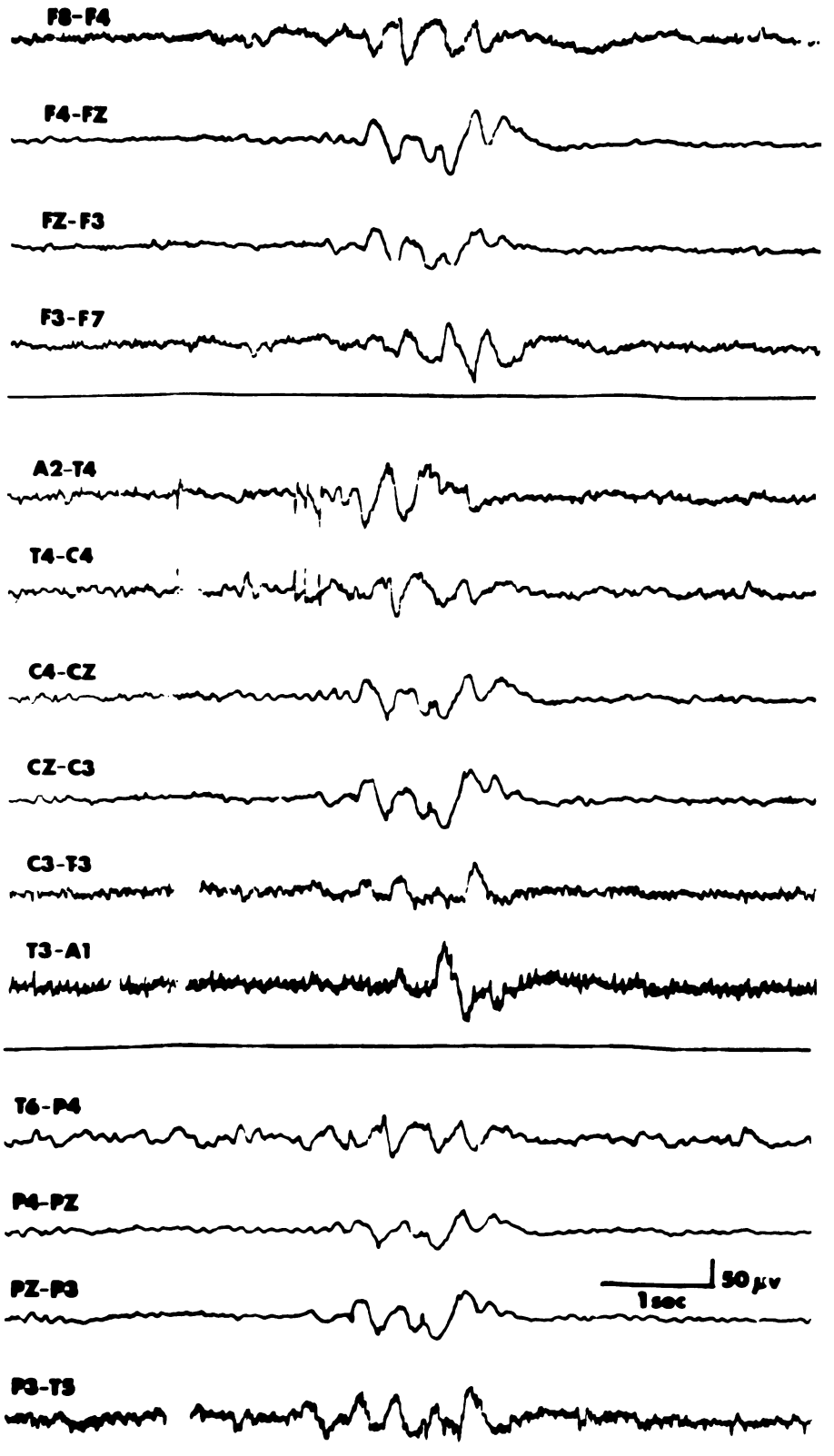

Figure 1 — EEG tracing of a 20 year old Friedreich's ataxia patient (Group I) showing bursts of medium to high voltage 3 c/s slow waves. Slightly irregular background activity. Similar bursts were found in some patients of group II.

poor performance, but Group II (I.Q.: 71.14) scored lower than Group I (I.Q.: 81.56) and, as shown above, had a greater incidence of EEG abnormalities. We have suggested (Bouchard et al., 1978) that the physical handicap does not explain the low nonverbal scores in the ARSACS patients (Group II) and this is supported by the fact that our Friedreich's ataxia patients were clinically more clumsy with their hands but performed better on the non-verbal scale. The I.Q. values shown above for each scale are not significantly different between the two groups, but the global I.Q. is significantly lower in Group II. (Group II: 78.27; Group I: 91.00; $p < .05$).

CONCLUSION

All the EEG abnormalities in our two groups of ataxic patients were mild and non specific, consisting of slow or irregular background activity, bursts of slow waves of sub-cortical origin, and poor driving response to ILS. This is in accordance with the previous analysis of EEG findings in Friedreich's ataxia (Guillain et al., 1942; Badiu and Popescu-Tismana, 1968; Davies, 1949 b; Rémillard et al., 1976). We consider that EEG is of little diagnostic value since no specific pattern could be identified in either group.

Quantitatively, the incidence of EEG abnormalities in our Group I (30%) is similar to that of the classical Friedreich's ataxia patients studied in 1976 by Rémillard et al., which is not surprising in view of the fact that these patients fit into the same clinical form of hereditary ataxia. ARSACS patients, however, have amuch greater incidence (63%) of EEG abnormalities and the findings are quite constant for a given patient. This is possibly related to the fact that the disease begins early in life at the time of brain maturation, even though it is not mainly a disease of the cerebral hemispheres. Nevertheless, the tendency of all patients of this group to show lower I.Q. performance, mainly in non-verbal tests, in addition to poorer EEG profile suggest more involvement of the cortical and sub-cortical structures than in the Friedreich's ataxia.

https://doi.org/10.1017/S0317167100119626 Published online by Cambridge University Press

We have no data of EEG's recorded in apparently normal individuals who developped the disease later and we do not have the EEG profile of unaffected siblings of this group. In Rémillard's study (1976), the EEG pattern of unaffected siblings was not significantly different from that of normal individuals. This gives little support to the hypothesis of genetic determination of electric abnormalities, at least for Rémillard's group, which is similar to our group I. On the other hand, some authors (Crighel and Ionasescu, 1962; Kissel et al., 1955) have described similar electric patterns in several members of "Friedreich families" among whom a few were clinically unimpaired; they suggest that the unaffected member with identical EEG could be a bearer of the disorder. In seven patients of our ARSACS group from three families, we have not been able to identify a familial electric pattern. The same thing can be said about our Friedreich group which included seven patients from three families, and in whom most of the EEG's were normal.

The Occipital driving response to ILS is poorer in both groups than in normals. This is clearly not a reliable finding because of its variability in the same patient, at least in ARSACS. Therefore, we conclude that driving responses are variable and inconsistent rather than abnormal in these patients.

There is no evidence in either group that EEG abnormalities progress as the disease goes on, and there is no significant difference with regard to age and sex. This is in accordance with the most recent study (Rémillard et al., 1976), and supports the view that EEG is of limited value in terms of prognosis.

Regardless of the groups, focal abnormalities were occasional, discrete, and were over the right hemisphere in four right handed patients and over the left hemisphere in one left handed patient. Although focal abnormalities do not necessarily mean functional disorder, this remains an interesting finding in view of the difficulties with handling non-verbal material in our patients. Davies (1949, a,b) has also reported right temporal slowing (case I and case II) in patients with behavior disorder but their handedness was not mentioned. Their I.Q. performance was slightly lower in the non-verbal test (Raven's progressive matrices) than in the verbal scales (Wechsler Bellevue scale). Comparison between Davies' group and ours as far as I.Q. is concerned needs careful interpretation since the psychological tests were not the same. In all our patients we have used the Ottawa-Wechsler intelligence battery for both verbal and non-verbal performance.

These are fragmentary data and more neuropsychological testings would be of interest in larger groups, with regard to EEG focal findings, memory, intelligence, behavior, right or left hemispheric dysfunction, and handedness, as has been studied in epileptic patients.

ACKNOWLEDGEMENTS

This work was supported by grants from l'Association canadienne de l'Ataxie de Friedreich. The authors wish to thank Dr. M. Filion for his advice and Mrs. A. Labrecque for her technical assistance.

REFERENCES

ANDERMANN, E., REMILLARD, G. M., GOYER, C., BLITZER, L., ANDERMANN, F. and BARBEAU, A. (1976). Genetic and family studies in Friedreich's ataxia. Can. J. Neurol. Sci., 3, 287-301.

BADIU, G. and POPESCU-TISMANA, G. (1968). Some electroencephalogrphic features of spino-cerebellar degeneration. Confin. Neurol., 30, 261-271.

BOUCHARD, J. P., BARBEAU, A., BOUCHARD, R. and BOUCHARD, R. W. (1978). Autosomal recessive spastic ataxia of Charlevoix-Saguenay. Can. J. Neurol. Sci., 5, 61-69.

CRIGHEL, E. and JONASESCU, H. (1962). Electroencephalographic findings in two families with Friedreich's ataxia. Confin. Neurol., 22, 28-39.

DAVIES, D. L. (1949 a). The intelligence of patients with Friedreich's ataxia. J. Neurol. neurosurg. Psychiat., 12, 34-38.

DAVIES, D. L. (1949 b). Psychiatric changes associated with Friedreich's ataxia. J. Neurol. Neurosurg. Psychiat., 12, 246-250.

GUILLAIN, G., BERTRAND, I., GODET, J. and GRUNER, J. (1942). L'électroencéphalogramme dans la maladie de Friedreich. C. R. Soc. Biol., 136, 494-495.

KISSEL, P., ARNOULD, G., HARTEMANN, P., DUREUX, J. and DEBRY, G. (1955). L'électroencéphalogramme dans la maladie de Friedreich. Rev. Neurol., 93, 761-764.

REMILLARD, G., ANDERMANN, F., BLITZER, L. and ANDERMANN, E. (1976). Electroencephalographic findings in Friedreich's ataxia. Can. J. Neurol. Sci., 3, 309-312.