# Electro-Retino-Encephalography in Leber's Tapeto-Retinal Dystrophy

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Leber's tapeto-retinal dystrophy, first described in 1869 by Theodor Leber, is classified among the diffuse dystrophies of the retina.[1-5]

Electro-retinography is always considered one of the main investigation methods for differential diagnosis of this type of retinal involvement. It is known that ERG is usually diminished or completely abolished in Leber's tapeto-retinal dystrophy, although this is not the case in many other similar fundus appearances, such as rubella retinitis, congenital syphilis, congenital optic atrophies, degenerative myopia, some macular involvements and Tay-Sachs disease. [2 3 5-15]

Some authors [2 12 16-19] have carried out combined ERG-EEG tests because they also indicated disturbances in the electric activities of the brain.

## *Materials and Methods*

This paper is part of a genetical, biostatistical and clinical investigation of Leber's tapeto-retinal dystrophy.[20] The study was made on 39 patients of different ages, the oldest being 30 years of age, and the youngest eight months old. Twenty-three of these patients were selected from the School for the Blind in Ankara, and the remainder from the outpatient Department of Ophthalmology, Hacettepe University Faculty of Medicine.

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Combined ERG-EEG investigations were possible on all the patients. Grass IV and Grass VI type electro-encephalographs were used for this purpose, one of the channels being devoted to ERG. Grass PS2 type photostimulators were used for light stimulus of 500,000 candle-light power and flashes of 10 millisecond duration were given 1-5 and 18 (Flicker) times per second, respectively.

The simple traces obtained with this ERG-EEG arrangement, though not very distinctive in detail, were compared with traces of a control group, and were found sufficient for differential diagnosis. In Figures 1, 2 and 3 normal electro-retinograms obtained from the control cases can be seen.

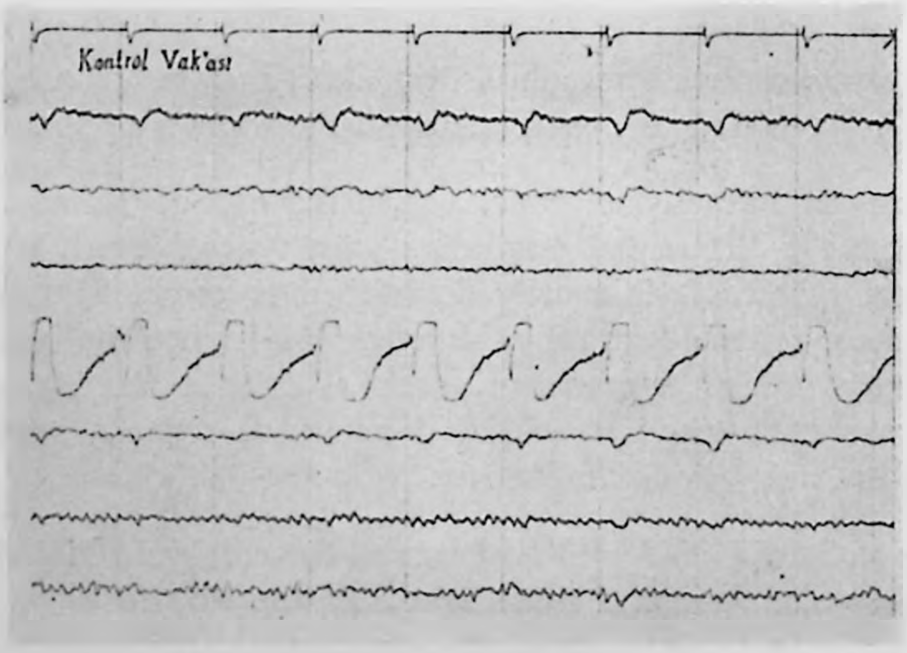


**Figure 1.** Electro-retino-encephalogram of a normal subject. 1 flash/second, 5th channel devoted to ERG.

*Results*

In 36 cases out of 39 the ERG was extinguished. Only three patients showed a subnormal and decreased response, the incidence of abolished ERG thus being 92.3 per cent. Figure 4 shows a subnormal response, and Figure 5 a completely extinguished ERG response in our series.

Twenty-six out of 39 EEG traces were normal. In 13 slight disturbances of background activity were seen over the parieto-occipital regions. No epileptiform disturbances were observed, although some authors[2 16] have recorded such attacks.

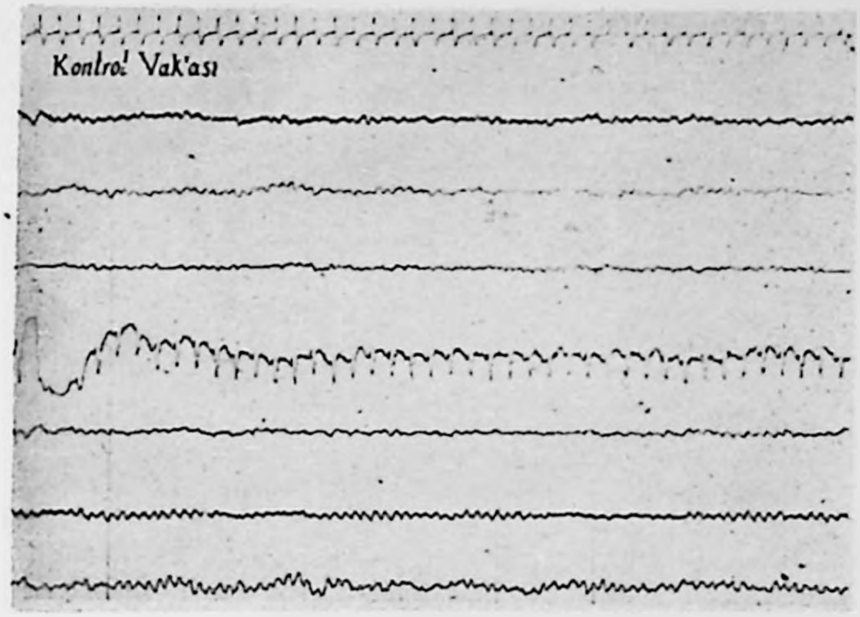


**Figure 2.** Electro-retino-encephalogram of a normal subject. 5 flashes/second, 5th channel devoted to ERG.

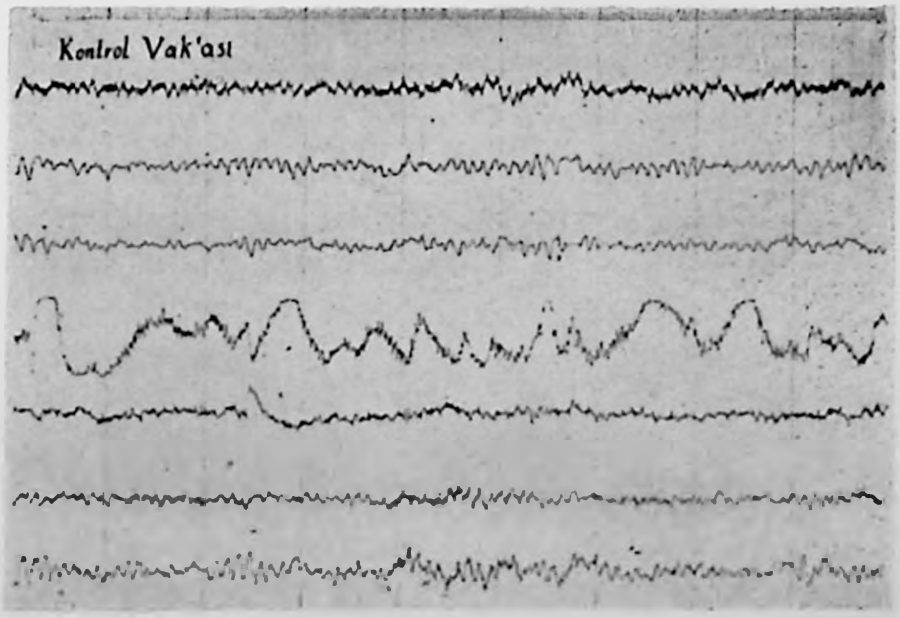


**Figure 3.** Electro-retino-encephalogram of a normal subject. 18 flashes (Flicker) per second, 5th channel devoted to ERG.

## *Discussion*

There seems to be a striking resemblance between the results found by other authors and our own, as can be seen in Table 1.

Disturbances in EEG do not represent a specific pathology or a cerebro-retinal pathology as far as we know.[20] [21]

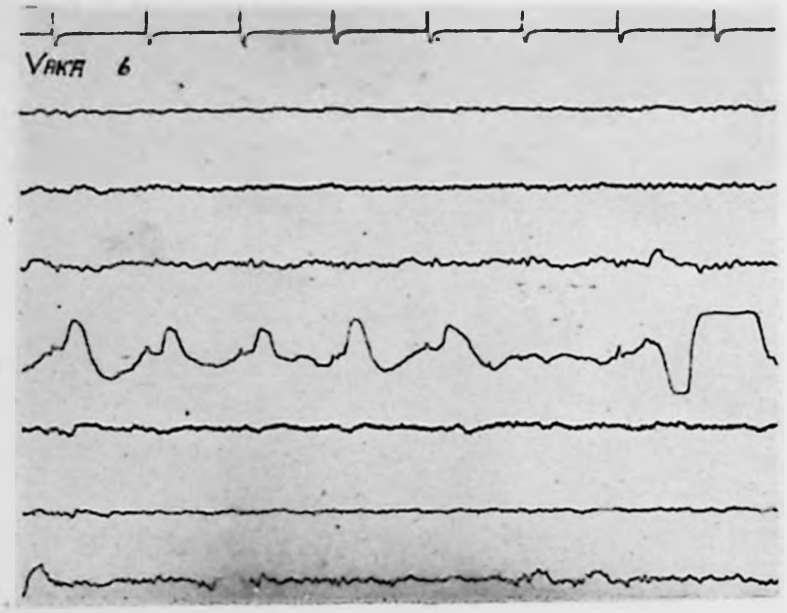


**Figure 4.** Subnormal ERG in Leber's tapeto-retinal dystrophy.

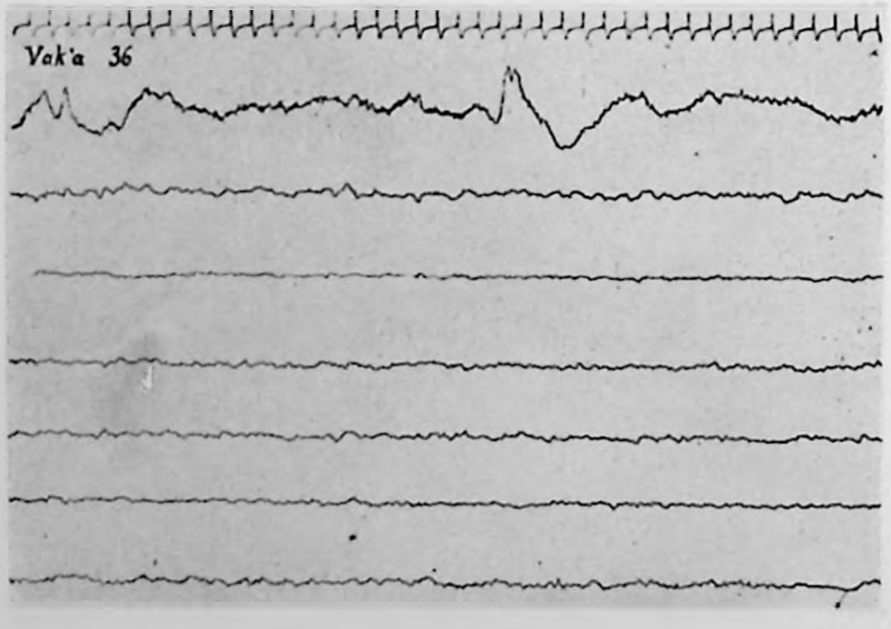


**Figure 5.** Completely abolished ERG in one of the cases of Leber's tapeto-retinal dystrophy.

TABLE I

| Other authors | % of Abolished ERG |
|---|---|
| Alström and Olson | 92 |
| Avanza | 100 |
| Schappert-Kimmijser et al | 100 |
| François and De Rouck | 72 |
| Ravault | 90 |
| Our series | 92.3 |

## TABLE II

Table 2 shows a comparison of our EEG findings with those of other authors

| Other authors | % of Disturbed EEG |
|---|---|
| Francheschetti-François-Babel | 42.8 (Epileptiform) |
| François-De Rouck | 10 (Epileptiform) |
| François-Stefens-De Rouck | 43 |
| Our series | 33.4 |

*Summary*

Electro-retinographic and electro-encephalographic investigations in Leber's tapeto-retinal dystrophy were carried out on 39 patients of different ages. In 92.3 per cent of the cases ERG was abolished, and in 33.4 per cent dysrythmic changes of the parieto-occipital type were present in EEG.

These results are similar to those of other authors, and emphasize once more the importance and value of ERG in the differential diagnosis of Leber's tapeto-retinal dystrophy.

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## *REFERENCES*

1. François, J.: Heredity in Ophthalmology, Mosby Co. St. Louis, 1961.
2. Franchescetti, A., François, J. and Babel J.: Les Hérédodégénérescences Chorio-Retiniénnes, Tome I, Masson et Cie, Paris, 1963.
3. Duke-Elder, Sir W. S.: System of Ophthalmology, Vol. X, Kimpton, London, 1967.
4. Waardenburg, P. J., Franchescetti, A. and Klein, D.: Genetics and Ophthalmology, Vol. I, Royal Van Gorcum Ltd., Assen, Netherlands, 1961.
5. Alström, C. H. and Olson, O.: Heredo-retinopathia congenitalis monohybrida recessiva autosomalis, Lund: Berlingska Boktryckeriet, Hereditas **43**: 1, 1957.
6. Avanza, C.: L'amaurosi o degenerazione tapeto-retinica congenita o infantile di Leber, Boll. Oculist **41**: 635, 1962.

7. Schappert-Kimmijser, J., Henkes, H. E. and van den Bosch, J.: Amaurosis congenita (Leber), Arch. Ophthal. 61: 211, 1959.
8. François, J.: The differential diagnosis of tapeto retinal degenerations, Arch. Ophthal. 59: 88, 1958.
9. Dieterle, P.: L'importance de l'électrorétinographie (ERG) pour le diagnostic différentiel entre la dégénéréscence tapéte-rétinienne primitive et secondaire, Opthalmologica 127: 557, 1954.
10. Straub, W.: Die bedutung des elektroretinogramms für die diagnostik der angeborenen tapeto-retinalen degeneration, Klin. Monats. Augenheilk, 134: 178, 1959.
11. François, J.: L'électro-rétinographie dans les dégénérescences tapéto-rétiniennes périphériques et centrales, Ann. Ocul. 185: 842, 1952.
12. François, J. and De Rouck, A.: L'intérêt de l'électrorétinographie dans le diagnostique de la cécité du nouveau-né, Ophthalmologica, 140: 1, 1960:
13. Dieterle, P.: L'importance de l'ERG pour le diagnostique différential entre la dégénérescence tapéto-rétinienne primitive et secondaire, Ophthalmologica, 127: 357, 1954.
14. Ravault, M. P.: L'atrophie optique de l'enfant, J. Méd. Lyon, 1100: 947, 1966.
15. Jayle, G. E., Boyer, R. L. and Saracco, J. B.: L'Electrorétinographie, Tome I-II, Masson et Cie., Paris, 1965.
16. François, J. and De Rouck, A.: Dégénéréscence tapéto-rétinienne congénitale de Leber, Bâsle: 18é Cong. Ass. Péd. Langue Fr. Karger, 1961.
17. François, J.: La dégénéréscence tapéto-rétinienne congénitale de Leber, Bull. Mém. Soc. Fr. d'Ophtal. 76: 1, 1963.
18. François, J., Stefens, R. and De Rouck, A.: L'électro-rétino encéphélographie dans la rétinopathie pigmentaire, Ann. Ocul. 187: 908, 1954.
19. Winkelmann, J. F. and Horsten, G.P.M.: Congenital blindness in the presence of a normal fundus, Ophthalmologica, 137: 423, 1959,
20. Fırat, T.: Clinical and genetical investigations in Leber's tapeto-retinal dystrophy, some characteristic aspects in Turkey (to be published), 1968.
21. Kalabay, O.: Personal Communication, 1968.