nodule tissulaire polylobé médiastinal antérieur mesurant 24 x 23,5 mm, siège de calcification centrale (Figure 1).

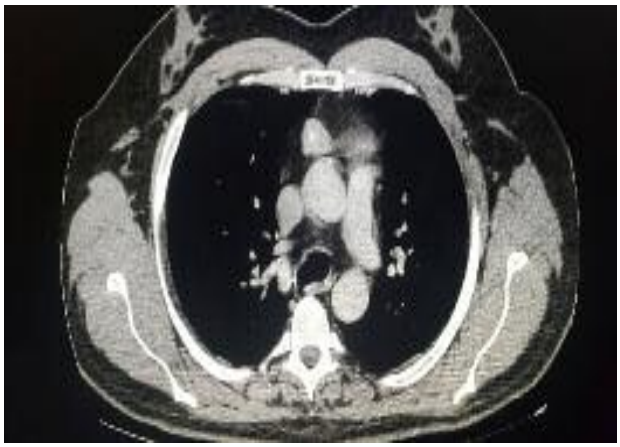

Figure 1: Scanner thoracique: nodule tissulaire polylobé médiastinal antérieur mesurant 24 x 23,5 mm, siège de calcification centrale.

La scintigraphie à l'octréoscan a mis en évidence un foyer d' hyperfixation médiastinal correspondant à un nodule calcifié de 25 mm sur le scanner de repérage.
Le patient a été donc opéré, il a eu une thymectomie totale emportant le nodule objectivé au scanner, et qui était situé au pole inférieur du lobe droit du thymus. L'examen anatomopathologique a conclu à une tumeur carcinoïde atypique du thymus, infiltrant la graisse médiastinale et le péricarde, stade III selon la classification de Masaoka modifiée. Les limites chirurgicales étaient saines. Un complément de traitement par radiothérapie a été indiqué.
L'évolution postopératoire était favorable avec disparition des signes d'hypercortisolisme.
La figure 2 représente l'évolution des taux de l'ACTH et du cortisol plasmatique avant et après thymectomie.

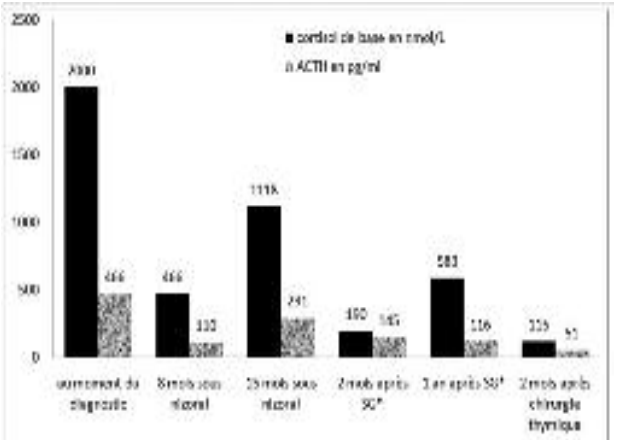

Figure 2 : Evolution des taux de l'ACTH et du cortisol plasmatique avant et après thymectomie.
*SG : surrénalectomie gauche

Conclusion

Le syndrome de cushing paranéoplasique secondaire à une tumeur carcinoïde thymique est une manifestation rare posant surtout le problème du diagnostic topographique. En effet, ces tumeurs sont de petite taille pouvant échapper aux techniques d'imagerie conventionnelle et même à la scintigraphie à l'octréotide marqué à l'Indium-111 (examen de référence) comme c'était le cas de notre patient. Dans cette situation, la tomographie par émission de positons au 18-fluorodeoxyglucose (TEP-^{18}FDG), qui n'était pas disponible dans notre pays, aurait été plus contributive.

Références

1. Bolland MJ, Holdaway IM, Berkeley JE et al. Mortality and morbidity in Cushing's syndrome in New Zealand. Clinical Endocrinology 2011; 75: 436–442.
2. Newell-Price J, Bertagna X, Grossman AB, Nieman LK. Cushing's syndrome. Lancet 2006; 367: 1605-17.
3. Isidori AM, Lenzi A. Ectopic ACTH syndrome. Arq Bras Endocrinol Metab 2007; 51 (suppl 8): 1217-25

Intrafamilial phenotypic variability in idiopathic Fahr's disease

Ben Amou Boutheina1, Kacem Imen2, Nasri Amira2, Ben Djebara Mouna2, Sidhom Youssef 2, Gargouri Amina2, Gouider Riadh1
1-Service de Médecine interne, Hôpital Mohamed Tahar Maamouri, Nabeul / FMT,
2-Service de Neurologie, UR12SP21,CHU Razi, La Manouba / FMT

Fahr's syndrome, or striato-pallido-dentate calcifications (SPDC), is a rare neurological disorder consisting in intracerebral calcifications. The idiopathic form defines the Fahr's disease: a rare familial idiopathic basal ganglia calcifications characterized by a highly genotypic and phenotypic polymorphism. It can be asymptomatic in thirty percent of cases (1). When it is symptomatic, three main phenotypes can be distinguished. Predominant cognitive and/or psychiatric signs; predominant extrapyramidal signs with parkinsonism and/or dystonia, and less frequently, predominant cerebellar ataxia (1-3). Clinical manifestations are not entirely correlated to morphological changes and functional abnormalities may precede morphological changes in the disease process (2-5). Fahr's disease can be sporadic or familial. The majority of reported familial cases demonstrates an autosomal dominant transmission (6,7).
Herein, we report on an autosomal recessive familial form of Fahr's disease with intrafamilial clinical heterogeneity.

Observations

Patient 1(IV-3)
The proband is a 43 year-old woman with a family history of an early death of a brother with a similar case. She was followed in the psychiatric department since she was 33 years old, for a schizophrenia-like psychosis and was treated by antipsychotic drugs. At the age of 40 years, she developed generalized seizures and was treated with Phenobarbital with a good progress. At the age of 42 years, she developed movement disorders. Neurological examination showed a mental retardation (IQ could not be estimated), quadripyramidal syndrome, a bilateral and symmetric extrapyramidal syndrome, choreic movements of the upper limbs and orofacial dyskinesia. Electroencephalography was normal. A brain CT scan and MRI revealed bilateral symmetric calcifications of basal ganglia and the cerebellum (Fig 2). The patient was seizure free under Phenobarbital, stabilized by antipsychotic treatment and keeps some orofacial dyskinesia.

Patient 2 (IV- 4)
The sister is 40 year-old woman. She has been under observation since the age of 20 years old in the psychiatric department for schizophrenia and mental retardation and was treated by anti-psychotics drugs. The neurological examination found a deep mental retardation and extrapyramidal syndrome with rigidity of the 4 limbs, a rest tremor of the upper limbs, and oral dyskinesia. A brain CT scan showed calcifications of basal ganglia (fig 3). She was stabilized by antipsychotic drugs.

Patient 3 (IV- 5)
The youngest brother; a 33 year-old man, had a history of febrile seizures at the ages of 5 and 10 years old. He had a milestone delay: he was hypotonic and began walking and talking at the age of 11. Neurological examination found mental retardation, behavioral disorders such as psychomotor agitation, spastic gait and generalized dystonia. The electroencephalography was normal. The brain CT scan and MRI were normal. He was treated with benzodiazepine, Phenobarbital and Baclofene. The

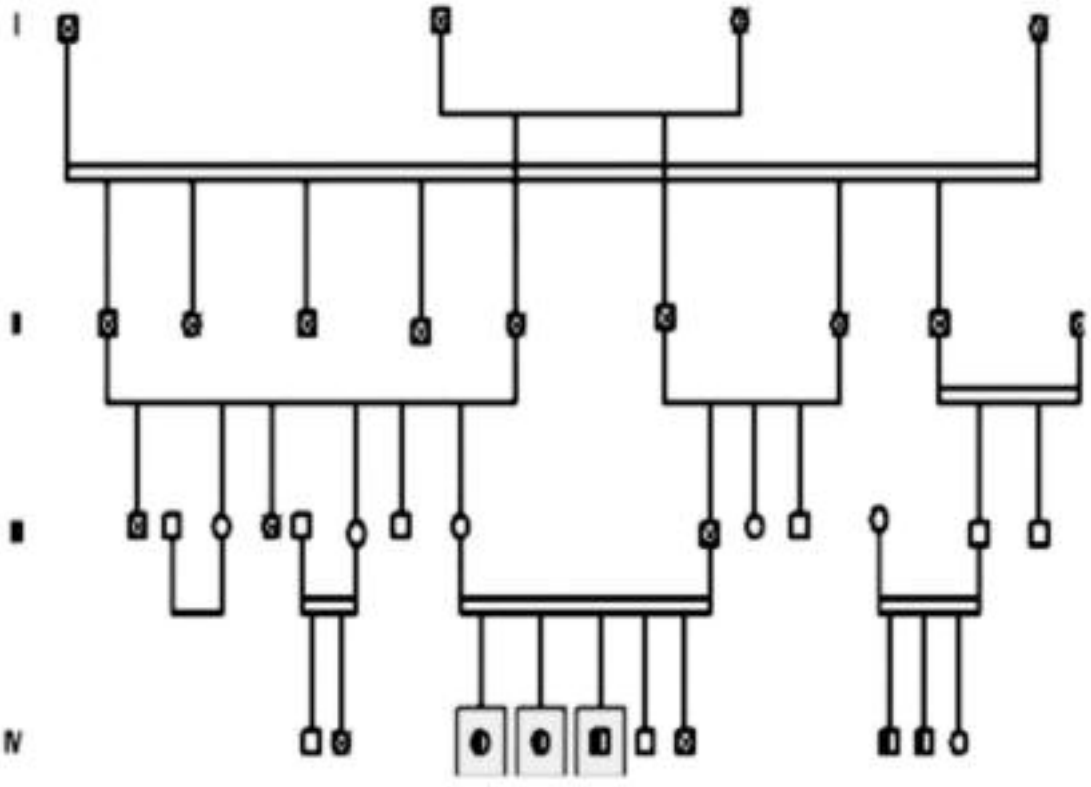

Figure 1: Family's Pedigree with inherited Fahr's disease. We report the case of three probands. Two other siblings have the same clinical manifestations, but they were not been available for study.

◐ Fahr's disease ▭ Our patients

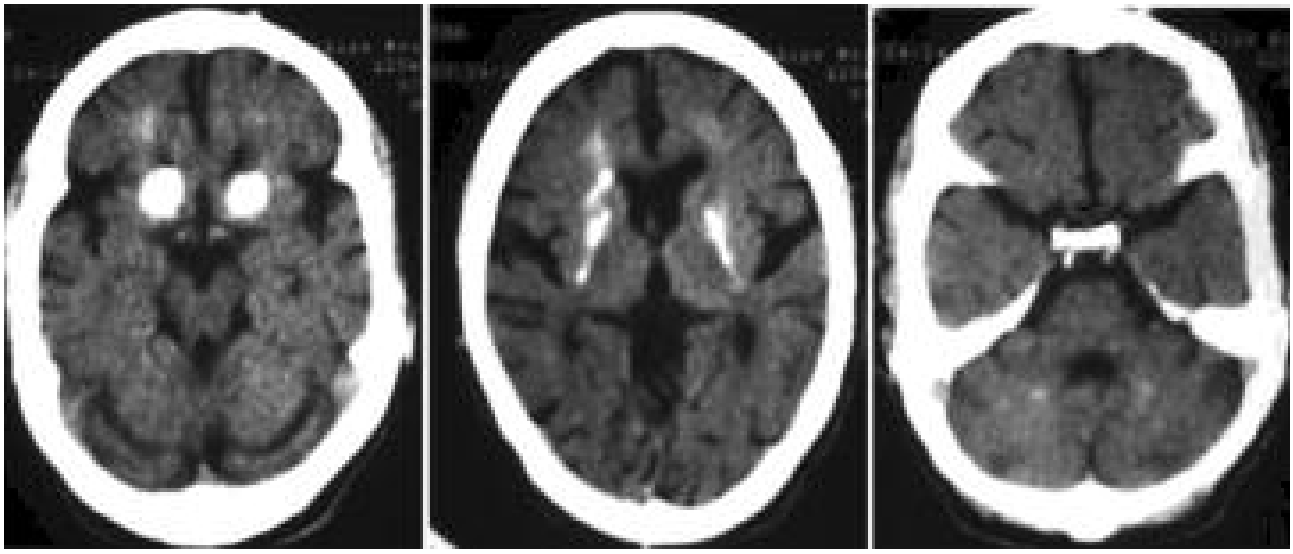

Figure2 : Brain CT scan of Patient IV-3showing calcifications of basal ganglia and the

patient was seizure free, but he still has generalized dystonia and behavioral disorders.

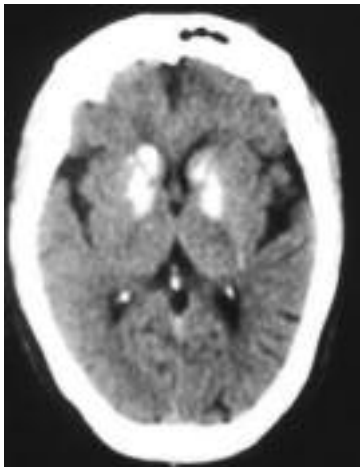

Figure 3: Brain CT scan of Patient IV-4 showing calcifications of basal ganglia

The mother denied any similar condition in the ascendant, she was asymptomatic with a normal neurological examination, and her Brain CT scan was also normal.

Conclusion

Herein we report a familial form of Fahr's disease with a likely Autosomal recessive transmission. Our family confirms the absence of parallelism and correlation between radiological and clinical finding and the clinical heterogeneity of the familial form of disease.

References

1. Ashtari F, Fatehi F. Fahr's disease: variable presentation in a family. Neurological Sciences 2010; 31:665-7.
2. Alemdar M, Selek A, Iseri P, Efendi H. Fahr's disease presenting with paroxysmal non-kinesigenic dyskinesia: a case report. Parkinsonism Related Disorders 2008; 14:69–71.
3. Casanova MF, Araque JM. Mineralization of the basal ganglia: implications for neuropsychiatry, pathology and neuroimaging. Psychiatry Research 2003; 12:159–87.
4. Hempel A, Henze M, Berghoff C, Garcia N, Ody R, Schroder J.PET findings and neuropsychological deficits in a case of Fahr's disease. Psychiatry Research 2001; 108: 133-40.
5. Le Ber I, Marié RM, Chabot B, Lalevée C, Defer GL. Neuropsychological and 18FDG-PET studies in a family with idiopathic basal ganglia calcifications. Journal of the Neurological Sciences 2007; 258: 115–22.
6. Wan-Jin Chen et al. Novel SLC20A2 mutations identified in southern Chinese patients with idiopathic basal ganglia. Gene 2013; 529: 159–62.
7. Dai X., et al. Identification of a novel genetic locus on chromosome 8p21.1-q11.23 for idiopathic basal ganglia calcification. American Journal of Medical Genetics Part B Neuropsychiatric genetics 2010 ; 153B : 1305–10

Ostéome ostéoïde du col du talus

Wafae Rachidi, Kawtar Nassar, Saadia Janani, Mohammed Rahmi, Abdelhak Garch, Ouafa Mkinsi.
Centre Hospitalier Universitaire Ibn Rochd / Université Hassan II/faculté de Médecine et de Pharmacie de Casablanca

L'ostéome ostéoïde est une tumeur ostéoblastique bénigne fréquente puisqu'il représente près de 10 % des tumeurs osseuses bénignes (1). Il a été décrit pour la première fois par Jaffe en 1935 (2). Il s'observe essentiellement entre 7 et 25 ans, avec une prédominance masculine (3). Il se localise avec prédilection au niveau des os longs, en particulier le fémur et le tibia qui représentent près de 50% des cas rapportés (4). L'atteinte du pied et de la cheville représente seulement 2 à 11% (2). Au pied, le talus est la