

# The Corpus Callosum Agenesis: A Case Report

Derbali F<sup>1,\*</sup>, Ben Fradj F<sup>2</sup>, Rezgui A<sup>2</sup>, Karmani M<sup>2</sup>, Hajji R<sup>1</sup>, Ben Abdallah O<sup>2</sup>, Mzabi A<sup>2</sup>, Laouani C<sup>2</sup>

<sup>1</sup>Internal Medicine Department - SidiBouزيد Regional Hospital –SidiBouزيد - Tunisia

<sup>2</sup>Internal Medicine Department - Sousse University Hospital – Sousse - Tunisia

\*Corresponding author: [derbalifatma@yahoo.com](mailto:derbalifatma@yahoo.com)

Received December 30, 2014; Revised January 16, 2015; Accepted January 18, 2015

**Abstract** Agenesis of corpus callosum is the most frequent cerebral malformations. Diagnosis is based on brain imaging. It is most often asymptomatic. Hyperhidrosis can be part of his clinical picture. We present the case of a 49 year old patient, unemployed, with a history of frontal lobe epilepsy and extra pyramidal syndrome. He consulted for a left hemiparesis with sweating attacks. Physical examination showed facial dysmorphism, psychomotor retardation, hemiparesis predominantly left brachiocephalic and bilateral pyramidal syndrome. Brain imaging showed, in addition to cerebral ischemia, a corpus callosum agenesis. This observation helped to highlight a rare disease whose clinical presentation is nonspecific. This malformation is to seek at prenatal, postnatal and even at the adulthood. Hyperhidrosis attacks without obvious underlying pathology should suggest this diagnosis.

**Keywords:** *corpus callosumagenesis, hyperhidrosis, brain malformations, epilepsy, brainimaging*

**Cite This Article:** Derbali F, Ben Fradj F, Rezgui A, Karmani M, Hajji R, Ben Abdallah O, Mzabi A, and Laouani C, "The Corpus Callosum Agenesis: A Case Report." *American Journal of Medical Case Reports*, vol. 3, no. 2 (2015): 27-29. doi: 10.12691/ajmcr-3-2-1.

## 1. Introduction

The corpus callosum (CC), the largest commissural in the brain, is essential for inter-hemispheric integration of sensory, motor, and higher-order cognitive information.

Corpus callosum agenesis (CCA) is the most frequent cerebral malformations. It can be symptomatic or asymptomatic. Diagnosis is based on imaging brain. Clinical signs are variable, they may include mental retardation, more or less moderate, seizures, "out control" behavior, facial dysmorphism and hyperhidrosis. Treatment is purely symptomatic.

We report the case of CCA revealed at adulthood by hyperhidrosis attacks.

## 2. Observation

A 49 year old man, unemployed, having the history of frontal lobe epilepsy, an extra-pyramidal syndrome consulted for a left hemiparesis installed three days ago with concept of sweating attacks. Physical examination objectified a broad forehead, hypertelorism, protruding tip of the nose, low-setears, psychomotor retardation, left hemiparesis and bilateral pyramidal syndrome. Brains canner objectified several old bilateral is chemicgaps, ischemia semi-recent at the territory of the right middle cerebral artery and total agenesis of the corpus callosum (Figure 1). We have not found other malformations. The karyotype was normal. Biological examination showed only hypercholesterolemia (8 mmol/ l) and hypertriglyceridemia (1.93 mmol/ l).The patient was treated with antiepileptics, antiplatelet agents and statins.

The evolution was marked by the disappearance of epileptic seizures, improvement of muscle strength but persistence of sweating attack.

## 3. Comments

Corpus callosum agenesis a brain malformation characterized by the absence of the main inter hemispheric commissure. Development of the corpus callosum (CC) occurs during weeks 6–20 of gestation, and development defects, observed in 0.3%–0.7% of individuals undergoing brain imaging, are among the most common of brain malformations [1]. It must be differentiated from other malformations (Dandy Walker disorder gyration, microcephaly, holoprosencephaly ...), abnormality chromosomal, metabolic disease (pyruvate deficit dehydrogenase), viral embryofetopathy and a product outlet toxic (drugs, alcohol) or drug (valproate ...). In our case, the diagnosis was confirmed by brain scanner which eliminated all other diagnosis. Kariotype didn't show abnormalities.

The CCA can be isolatedor belong malformation syndrome. Indeed, there are many syndromes with an "associated" CCA as Aicardisyndrome, orofacial digital, Anderman, Shapiro. This latter includes hyperhidrosis attacks and hypothermia with CCA [2]. Our case associate also CCA and hyperhidrosis.

Hypothalamic thermostat disorderis the cause of hypothermia. The mechanism that lead to isolated agenesis of the corpus callosum still unknown. An anomaly of the commissural plate, defect migration callous axons or an abnormality of the corpus callosum neurons have been suggested [3]. Isolated CCA can be

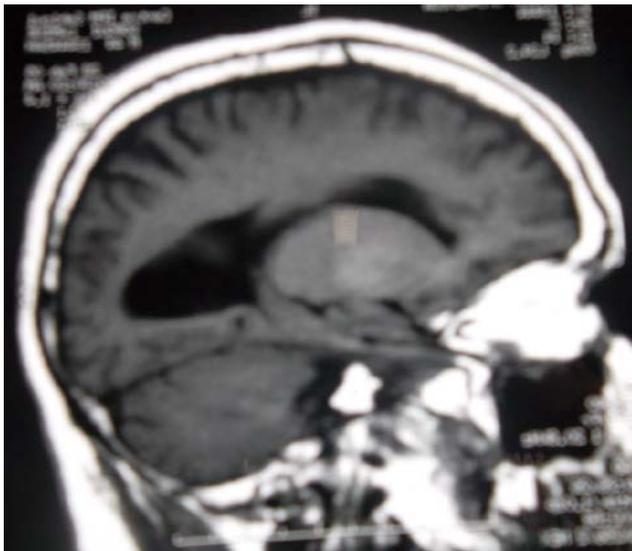
sporadic, X-linked recessive, autosomal dominant or recessive [4]. If several chromosomes seem involved in the development and maintenance of the corpus callosum (1, 8, 13, 15, 18, 21, X ...), no gene has been identified for these isolated agenesis.

In Tunisia a genetic study was performed on 8 families has concluded to heterogeneity of genetic profile of subjects having a thin CC [5]. In 50% of cases, the CCA is isolated. It may be symptomatic or asymptomatic. Our patient has an isolated and symptomatic form. Indeed, we found no associated malformation. The main clinical signs was epilepsy. Cerebral ischemia was anepi-phenomenon. The patient had several cardiovascular risk factors. The mental retardation is usually detected at the time of schooling. The intelligence quotient (IQ) testis normal in 2/3 of cases, there may be a moderate to severe intellectual deficit in 1/3 of cases [6,7]

Epilepsys found in approximately 50% of patients and consists of partial or generalized seizures. It is often easy to control. Several authors are interested in studying the relationship between epilepsy and abnormal body callosum. As was the case of our patient, partial epilepsys the most frequent form [8,9,10].

Behaviours problems are sometimes autistic type and emotional disturbance is accompanied by, a lack of attention or hyperactivity [11]. Taylor and David have 28 types of objective disorder Mental among 55 patients with CCA [12].

Dysmorphic, when it exists, is secondary to dilation accompanying ventricular malformation: hyper-telorism, protruding bumps frontal and macrocranium [9]. Our patient had objectified a broad front, hyper-telorism, protruding nose and low-set ears. Bilateral pyramidal syndrome, when present, is often integrated within a subtype called "Andermann syndrome" [13,14]. Some observations of spastic paralysis have been reported. Our patient has an extra pyramidal syndrome. Asymptomatic forms exist and the diagnosis is often incidental [15].



**Figure 1.** Total agenesis of the corpus callosum; MRI findings

Diagnosis is based on brain imaging (ultrasound transfontanellar, computed tomography, magnetic resonance imaging (MRI)) [16]. There is a dilation of the occipital horns (colpocéphalie); the corpus callosum is not viewed. Brain imaging suggest the absence of other

anomalies. In our case, diagnosis was made by brain scanner and MRI (Figure 1). Antenatal diagnosis of the malformation is now commonly performed by antenatal ultrasound and MRI. It is, however, difficult to give a prenatal counsel [14].

Treatment is symptomatic: Anti-epileptic treatment in case of seizures, psychomotor support, speech therapy, psychotherapy if psychomotor disorder [9]. Our patient remained table for a long time thanks to anti-epileptics. The diagnosis of CCA was made on the occasion of hyperhidrosis attacks and cerebral ischemia.

## 4. Conclusion

The CCA is a common malformation. It may be asymptomatic or revealed by a highly polymorphic and non-specific clinical feature. It should be researched in prenatal, postnatal and even in adulthood. Hyperhidrosis attacks without obvious underlying pathology should consider this diagnosis.

## References

- [1] Paul, L.K. Developmental malformation of the corpus callosum: a review of typical callosal development and examples of developmental disorders with callosal involvement. *J. Neurodev Disord* 2011; 3: 3-27.
- [2] Shenoy C. "Shapiro syndrome". *QJM*. 2008; 101 (1): 61-2.
- [3] Schell-Apacik CC, Wagner K, Bihler M, Ertl-Wagner B, Heinrich U, Klopocki E, et al. Agenesis and Dysgenesis of the Corpus Callosum: Clinical, Genetic and Neuroimaging Findings in a Series of 41 Patients. *Am J Med Genet*. 2008; 146A: 2501-11.
- [4] Boland E, Clayton-Smith J, Woo VG, McKee S, Manson FD, Medne L, et al. Mapping of deletion and translocation breakpoints in 1q44 implicate the serine/threonine kinase AKT3 in postnatal microcephaly and agenesis of the corpus callosum. *Am J Hum Genet*. 2007; 81 (2): 292-303.
- [5] Boukhris A, Stevanin G, Feki I, Denis E, Elleuch N, Miladi MI et al. Hereditary Spastic Paraplegia With Mental Impairment and Thin Corpus Callosum in Tunisia. *Arch Neurol* 2008; 65 (3): 393-402.
- [6] Brown WS, Paul LK, Symington M, Dietrich R. Comprehension of humor in primary agenesis of the corpus callosum. *Neuropsychologia* 2005; 43: 906-16.
- [7] Panos PT, Porter SS, Panos AJ, Gaies RN, Erdberg PS. An evaluation of a case of agenesis of the corpus callosum with Rourke's nonverbal learning disorder model. *Arch Clin Neuropsychol*. 2001; 16 (5):507-21
- [8] Khanna S, Chugani HT, Messa C et Curran JG. Corpus callosum agenesis and epilepsy: PET findings. *Pediatr Neurol* 1994; 10 (3): 221-7.
- [9] Grewal HK, Almulhassani A, Grewal J, Slater JD. Partial agenesis of the corpus callosum in a patient with juvenile myoclonic epilepsy. *Epileptic Disord* 2007; 9 (4): 461-4.
- [10] Dubravka ŠG, Antonija RB, Klementina R, Tanja G, Elizabeta DH. Neurological and psychiatric aspects of corpus callosum genesis. *Psychiatria Danubina* 2010; 22 (2): 282-5.
- [11] Devinsky O, LaffR. Callosal. Lesions and behavior: history and modern concepts. *Epilepsy & Behavior* 2003; 4 (6): 607-17.
- [12] Taylor M, David AS. Agenesis of the corpus callosum. A United Kingdom series of 56 cases. *J Neurol Neurosurg Psychiatry* 1998; 64 (1): 131-4.
- [13] Dupré N, Howard HC, Mathieu J, Karpati G, Vanasse M, Bouchard JP et al. Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. *Ann Neurol* 2003; 54 (1): 9-18.
- [14] Conti V, Marini C, Gana S, Sudi J, Dobyns WB, Guerrini R. Corpus Callosum Agenesis, Severe Mental Retardation, Epilepsy, and Dyskinetic Quadriparesis Due to a Novel Mutation in the Homeodomain of ARX. *American Journal of Medical Genetics* 2011; 155A (4): 892-7

- [15] Moutard ML, Kieffer V, Feingold J, Kieffer F, Lewin F, Adamsbaum C et al. Agenesis of corpus callosum: prenatal diagnosis and prognosis. *Childs Nerv Syst* 2003; 19 (7-8):471-6.
- [16] Sainthouse AM, Fytche DH, Howard RJ, Williams SC, Stewart AL, Rooney M et al. The functional significance of perinatal corpus callosum damage: fMRI study in young adults. *Brain* 2002; 125 (pt 8): 1782-92.