

Research Article

# Epidemiological and Clinical Study of Familial Epilepsy in Lubumbashi

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## Abstract

Epilepsies are very ancient diseases and the most widespread neurological diseases in the world. They are common conditions that constitute a heterogeneous group from an etiological perspective. The annual incidence of epilepsy is estimated at around 50 new cases per 100,000 inhabitants per year in industrialized countries, while in developing countries, this figure is higher, ranging from 100 to 190 new cases per 100,000 inhabitants per year. We conducted a cross-sectional analytical prospective study with stratified probabilistic sampling to select the subjects who participated. A total of 72 families were collected and subjected to statistical analyses. The patients living with epilepsy were aged 40 years or younger and had a family history of epilepsy. We found that the prevalence of familial epilepsy in Lubumbashi is 11.23% of cases and the mean age of patients with epilepsy and a family history of epilepsy is  $15.24 \pm 11.03$  years, with a median of 14 years, a 1st quartile of 7 years and a 3rd quartile of 20 years. The sex ratio is 1.32 in favor of men and generalized tonic-clonic seizures represent 70.83% of cases, followed by absence seizures at 8.33%. The frequency of other seizures fluctuates between 1.39% and 4.17% of cases.

**Keywords:** Epidemiology; Clinical; Epilepsy; Familial; Lubumbashi

## Introduction

Epilepsies, very ancient and the most widespread neurological diseases in the world, are frequent conditions constituting a heterogeneous set from an etiological point of view. Most

cases are multifactorial pathologies whose determinism involves environmental and genetic factors, which depend on the group of epilepsies considered. This means that these diseases represent a source of tension between magical and scientific conceptions, between superstitious beliefs and rational explanations [1,2].

## Incidence and Prevalence

The annual incidence of epilepsy is estimated at approximately 50 new cases per 100,000 inhabitants/year in industrialized countries, while in developing countries, this figure is higher, ranging from 100 to 190 new cases per 100,000 inhabitants/year [3]. Among the reasons that can explain this difference in incidence are the high risk of cerebral infections (neurocysticercosis, meningitis, malaria), obstetric complications and malnutrition [4]. In the Arab world, the average incidence of epilepsy was estimated through a study at approximately 56 new cases/100,000 inhabitants/year [5].

According to studies carried out worldwide, the average prevalence of epilepsy is estimated at approximately 8.2 per 1000 in the

general population [6,7]. However, this could be an underestimation, as some studies conducted in developing countries (Colombia, Ecuador, India, Liberia, Nigeria, Panama, United Republic of Tanzania and Venezuela) suggest a higher prevalence, greater than 10 per 1000 [8-11].

A multicentric cross-sectional study was conducted in 2012 in Algeria, with the aim of determining the national prevalence of epilepsy. This study included 8,046 subjects aged over two months, distributed across five regions of the country [12-14]. Sixty-seven patients were identified as having active epilepsy, giving a crude prevalence ratio of 8.32 per 1000 (95% CI: 6.34-10.3) and an age-adjusted ratio of 8.9 per 1000. The highest ratio (16.92 per 1000) was noted in the 10-19 age group [15]. Bora, et al., 2015, in their study on the epidemiology, risk factors and treatment of people living with epilepsy, noted a hospital prevalence of 11.9% of cases. This frequency is very high compared to other studies.

### *Mortality*

It has been shown in one study that patients suffering from epilepsy had a mortality rate 2 to 3 times higher than that of the general population [16-20]. This excess mortality was mainly noted in patients who suffered from pharmacoresistant partial epilepsy, where the standardized mortality ratio was 2 to 3 times higher than that of the general population [21]. Half of the deaths affecting this population are directly linked to the occurrence of epileptic seizures, through accidents, status epilepticus or Sudden Unexpected Deaths (SUDEP) [22].

The frequency of SUDEP deaths was estimated at 0.35 per 1000 person-years in an unselected population of epileptic subjects [23]. In patients with refractory epilepsy, the annual risk of SUDEP was estimated at 0.4%. This risk increases with the frequency of generalized tonic-clonic seizures, the absence of antiepileptic treatment reflecting poor adherence or, conversely, polytherapy, an indirect indicator of disease severity, an early age of onset or a prolonged duration of epilepsy [24-28].

Furthermore, classical data consider that approximately 10% of deaths in epilepsy can be attributed to suicide that the suicide rate is five times higher in people with epilepsy compared to the general population and that it could be multiplied by 25 following epilepsy surgery [29-31]. Idiopathic Generalized Epilepsies (IGEs) are a common group of epilepsies with a good prognosis and represent 15 to 20% of epilepsy cases. They are characterized by generalized epileptic seizures, including absence seizures, myoclonic seizures, generalized tonic-clonic seizures and myoclonic-tonic-clonic seizures. EEG can show paroxysmal patterns such as generalized spikes/polyspikes-slow waves. Generally, IGEs include four syndromes: Childhood Absence Epilepsy (CAE), Juvenile Absence Epilepsy (JAE), Juvenile Myoclonic Epilepsy (JME) and epilepsy with Generalized Tonic-Clonic Seizures Alone (GTCA) [32].

Studies by Sheidley BR, et al., and Truty R, et al., showed an increase in cases of genetic epilepsies for which specific treatment or a change in management is indicated. Advances in genetic testing over the past decade have led to a rapid increase in the understanding of the genetic basis of epilepsy [33]. Although the first genes associated with epilepsy were identified in 1995, few causal or susceptibility variants were confirmed until the past decade. The emergence of arrays and Next-Generation Sequencing (NGS) technologies has led to the identification of hundreds of variants and genes associated with epilepsy. There is broad consensus that the majority of otherwise unexplained epilepsies are due at least in part to genetic factors. Thus, genetic testing for patients suffering from epilepsy has also evolved to include many clinically available testing options, including genome-wide Comparative Genomic Hybridization/Chromosomal Microarray (CGH/CMA), Multi-Gene Panel (MGP), Exome Sequencing (ES) and Genome sequencing (GS). These tests are used to determine copy numbers and sequence variants involving epilepsy-related genes [34].

Studies by Marini C, et al., 2004 and Vadlamudi L, et al., 2014, demonstrated that genetic factors play important roles in the pathogenesis of IGEs with complex inheritance. In Algeria, in her work on the search for genetic susceptibility variants to epilepsy in Algerian families, Amina Chentouf (2017) showed that among sixty-five epileptic families, the average age of onset of the disease was 9.5pm 6.1 years, with a slight male predominance (sex ratio: 1.35) [35]. Generalized seizures were slightly more frequent than focal seizures (50% vs. 40%) with a parental consanguinity rate of 50%. Phenotypic concordance was observed in 2/3 of families. Based on pedigree analysis, epilepsy was transmitted in an Autosomal Dominant (AD) mode in 29 families (44.6%) and in an Autosomal Recessive (AR) mode in 23 families (35.4%). Genetic analyses identified mutations in the EPM1 gene in patients with progressive myoclonic epilepsy and a mutation in the RELN gene in individuals with temporal lobe

epilepsy.

M. Boanimbek Bakoume Bernard Baudouin, in his study on the etiologies of epilepsies in the neurology department of the Marrakech university hospital, found that initially generalized seizures were present in 80 patients in his study and represented 26.67% of seizures and that tonic-clonic seizures were the most common type of generalized seizure, accounting for 57.5% of generalized seizures alone [36]. Myoclonus was present in 16 patients or 20% of generalized seizures, while absence seizures were present in 14 of his patients or 17.5%. Atonic seizures were found in 4 patients or 5% of generalized seizures.

In their study in Pointe Noire on the Clinical, Etiological and Therapeutic Aspects of Infantile and Childhood Epilepsy, Sounga Bandzouzi, et al., found that thirty-two (82.1%) infants and children had idiopathic generalized epilepsy, of which 28 (87.5%) had epilepsy with generalized tonic-clonic seizures and four (12.5%) had absence epilepsy [37]. Of the seven infants and children with focal epilepsy, five (71.4%) had epilepsy with centro-temporal spikes (BECT); while epilepsy with frontal seizures was observed in two participants (28.6%).

It was reported in 2023 that nearly 80% of the 50 million people living with epilepsy live in low-income countries, including the Democratic Republic of Congo (DRC) and this situation represents a heavy health and socio-economic burden because gaps in etiological diagnosis and therapeutic coverage exceed 75%. Therefore, although several studies on epidemiological and clinical aspects have been carried out, as shown by this literature review, it is still important to deepen this study to determine the specificities in our environments because we believe that the African population still hides a vast diversity that could result in phenotypic variability compared to other populations.

Ntenga P, et al., in their study on epilepsy and schooling rates in Congolese children, showed that in 41% of cases, children living with epilepsy had a family history of epilepsy and that their average age was 9.6pm3.9 years with an average age of seizure onset of 5.8pm3.0 years. This percentage of family history of epilepsy, i.e., 41% of cases, could suggest a genetic character in our environment. Thus, our objective in this study was to determine the epidemio-clinical characteristics of familial epilepsy in Lubumbashi. This work aims to open a path to the study of epilepsy genetics in Lubumbashi with the goal of identifying and characterizing new epilepsy genes in Lubumbashi.

## **Methodology**

### *Study Setting and Location*

Our investigations are carried out at the Joseph Guislain Brothers of Charity Neuropsychiatric Center in Lubumbashi, at the University Clinics of Lubumbashi and at the Jason Sendwe Provincial General Reference Hospital in Lubumbashi. The Dr. Joseph Guislain Neuropsychiatric Center, ASBL / Brothers of Charity, is one of the specialized health structures in the Haut-Katanga province that cares for mentally ill patients. This center is located in the Kimbeimbe district, Joli site cell, on the Likasi road in the city of Lubumbashi, Haut-Katanga province. It receives all patients from the Haut-Katanga province with neuropsychic problems, as well as those from neighboring provinces and neighboring countries such as Zambia and Angola. It is located in the Lubumbashi health zone and in the Kimbeimbe health area [38]. The Jason SENDWE provincial reference hospital is located in the Democratic Republic of Congo, Haut-Katanga province, city of Lubumbashi. It is bordered: to the east by Sendwe Avenue separating it from the provincial tax office, to the west by the health division and the assistants' camp, to the north by Likasi Avenue separating it from the commercial district and to the south by the Sisters of Charity house and Wema high school. The University Clinics of Lubumbashi, which are located in the city and health zone of Lubumbashi, are bordered to the north by KAMBOVE Avenue, to the south by NDJAMENA Avenue, to the East by KASAI Avenue and to the west by TUENDELEE Avenue.

### *Study Type*

This is a cross-sectional analytical study conducted from October 2020 to October 2024, which included epilepsy cases presenting for consultation during the study period.

### *Study Population*

Patients aged 40 years or less presenting with epilepsy and their families received at the aforementioned investigation sites, with a notion of familial epilepsy discovered or reported during consultation, will be considered as potential candidates for this study.

**2.4. Sample** We performed stratified probabilistic sampling during this study for the selection of participants.

*Inclusion Criteria:* Patients aged 40 years or less with epilepsy who signed the consent and/or assent to participate were included in this study.

Must be of Black race and Congolese nationality (not of mixed race).

*Exclusion Criteria:* Genetic pathologies in which epilepsy is not the essential symptom, such as trisomy 21, were excluded from this study.

#### *Data Collection Material*

Data was collected and stored on data collection forms. At the end of each week, we systematically checked all completed forms to ensure they were properly filled out and to correct any errors on the same day or the next day by re-consulting the files or registers. The family tree (pedigree) was established for the patient to determine the recessive (if there is a skip generation) or dominant (disease present in each generation) characteristic.

#### *Study Parameters Sociodemographic Characteristics*

Sex: male (M), female (F) Age (date of birth: day/month/year)

#### *Clinical Parameters*

Age of seizure onset (in years)

Time elapsed between the onset of epileptic seizures and the date of consultation (in years) Family history of epilepsy: brother/sister, parents, grandparent, paternal/maternal side.

Clinical form of epileptic seizure:

Generalized seizure: GTC, atonic seizure, tonic seizure, myoclonic seizure, absence seizure Focal seizure: partial (without alteration of consciousness), complex (with alteration of consciousness)

#### **Therapeutic Parameters**

##### *Therapeutic Protocol*

*Monotherapy:* administration of a single antiepileptic drug

*Bitherapy:* administration of 2 antiepileptic drugs

*Tritherapy:* administration of 3 antiepileptic drugs

*Drugs used:* Valproic acid Carbamazepine Phenobarbital, Dihydan, levetiracetam, pregabalin, other

*Paraclinical parameters EEG:* search for paroxysmal grapho-elements (spike-wave, polyspike, polyspike-wave, slow waves notched synchronous spikes)

#### *Statistical Studies*

Data was collected and entered using Epi Info™ version 7.2.1.0 and SPSS software. Univariate analysis allowed us to calculate the frequency in terms of percentage for qualitative variables. The calculation of the mean, median and standard deviation allowed us to analyze quantitative variables. Bivariate analysis allowed us to compare means according to the distribution of sociodemographic, clinical and paraclinical characteristics. The chi-square test was performed. The Odds Ratio (OR) with a 95% Confidence Interval (CI) and a significance was set for  $OR > 1$  and a  $p < 0.01$  in order to look for the link between the variables.

#### *Ethical Aspects*

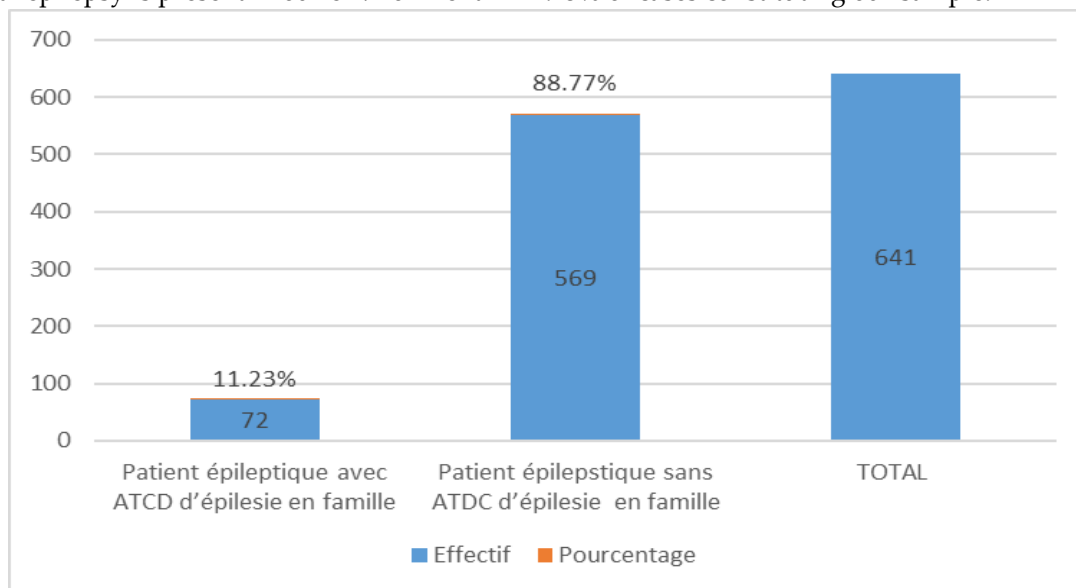
We took ethical aspects into consideration during this research. Indeed, the collection and analysis of data were subject to full confidentiality. In addition, the opinion of the study participants was sought and respected directly (adults) or indirectly (parents and/or accompanying children).

#### **Results**

The results of the present study concern both the general characteristics of the sample and those of the family tree. The first part concerns the prevalence of familial epilepsy in Lubumbashi, the average age of patients and their sex. General characteristics of the sample. I.1 Prevalence

This Fig. 1, originally a bar chart, would show the percentage of familial epilepsy cases within the sample. It appears from this

figure that familial epilepsy is present in our environment in 11.23% of cases constituting our sample.



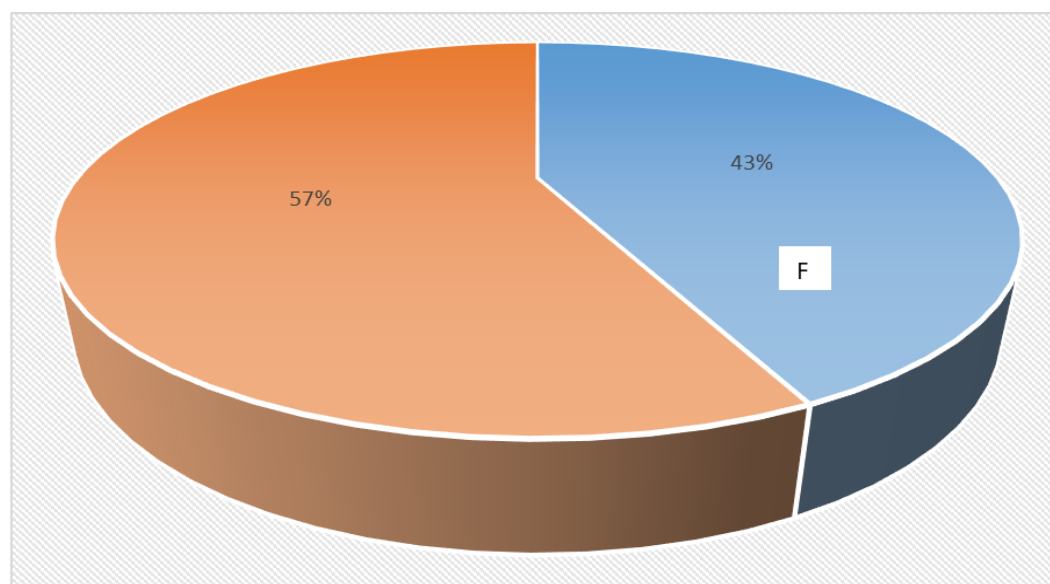
**Figure 1:** Prevalence of familial epilepsy.

Age and Sex Table 1. Distribution of patients by mean age The mean age and median of patients with a history of familial epilepsy are shown in the following table.

Age Moyen Des Patients			
Obs	Mean		Std Dev
72	15,24 ans		11,03
Median			Mode
25%	14 ans	75 %	17 Ans
7 years		20 years	

**Table 1:** Mean age of patients.

It appears from this table that the patients had a mean age of 15.24pm11.03 years, with a median of 14 years, 7 years at the 1st quartile and 20 years at the 3rd quartile. The distribution of patients according to their sex is shown in the Fig. 2 below. It emerges from this figure that the sex ratio is 1.32, in favor of men.



**Figure 2:** Distribution of patients by sex.**Distribution according to other parameters**

Table 2-5 and Fig. 3- 7 present other types of patient distribution.

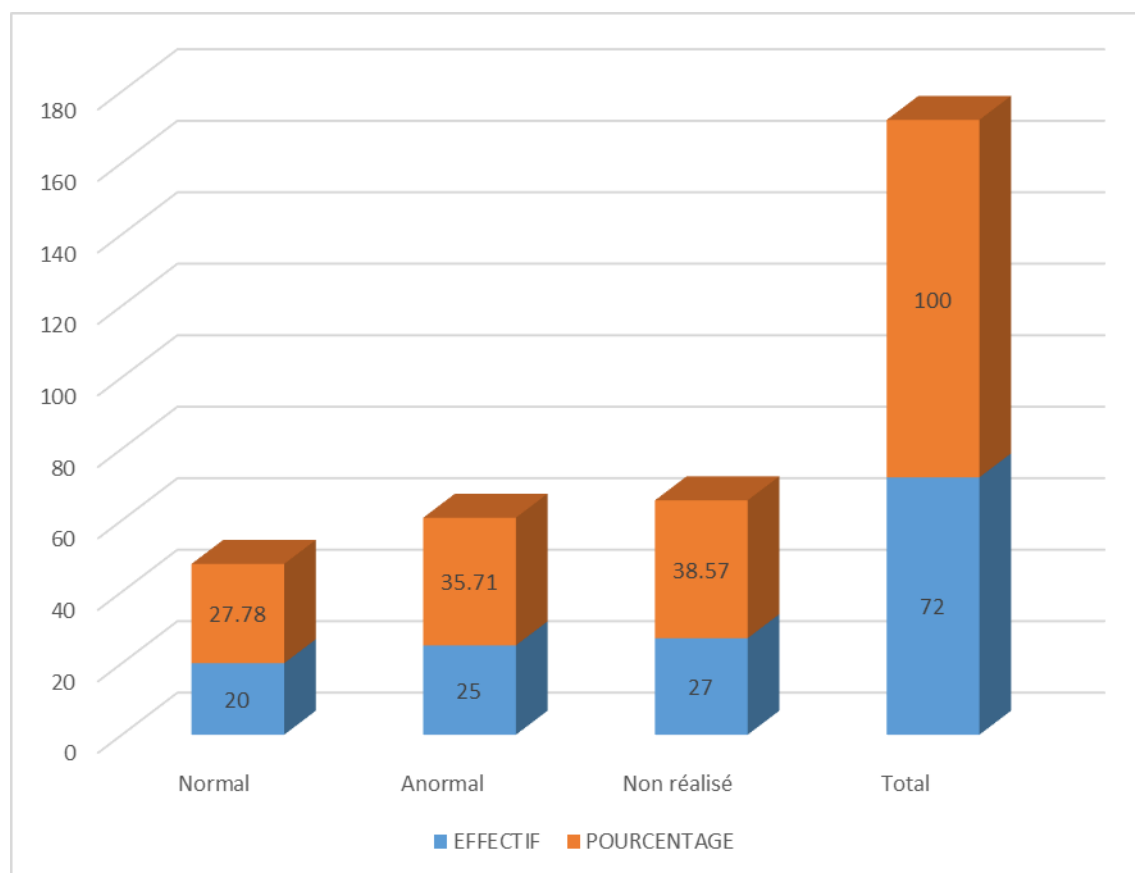
Type of Seizure	n	%
Partial seizure secondarily generalized	3	4,17%
Complex partial seizure	3	4,17
Myoclonic	3	4,17
Atonic seizure	3	4,17
Tonic seizure	1	1,39
Absence seizure	6	8,33%
Generalized tonic-clonic seizure	51	70,83%
Simple partial seizure	2	2,78%
Total	72	100

**Table 2:** Distribution of patients according to seizure types.

We observe from this table that generalized tonic-clonic seizures were the most common with 70.83% of cases, followed by absence seizures with 8.33% of cases. The frequency of other seizures ranged between 1.39% and 4.17% of cases. The EEG results in patients with a family history of epilepsy are shown in Fig. 3 below.

*a) Distribution of patients according to EEG results*

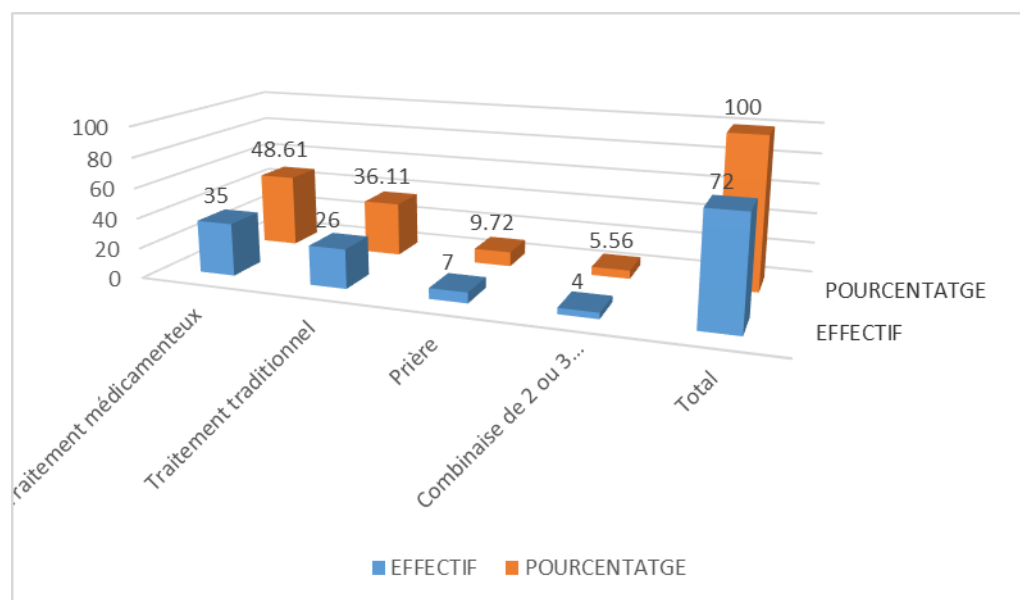
This Fig. 3, originally a bar chart, would illustrate the proportion of patients whose EEG was performed, normal or abnormal. Our results show that EEG was performed in the majority of patients (63.49% of cases) and was normal in 27.78% of cases and abnormal (presence of paroxysmal grapho-elements) in 35.71% of cases. The non-performance of this examination represented 38.57%.

**Figure 3:** According to EEG results.

Furthermore, the results of the patients' therapeutic journey, shown in Fig. 4, indicate that in 48.61% of cases, patients had followed a drug treatment before arriving at the hospital and that traditional treatment (36.11%) and prayer (9.72%) accounted for a total proportion of 45.83%.

*b) According to therapeutic itineraries*

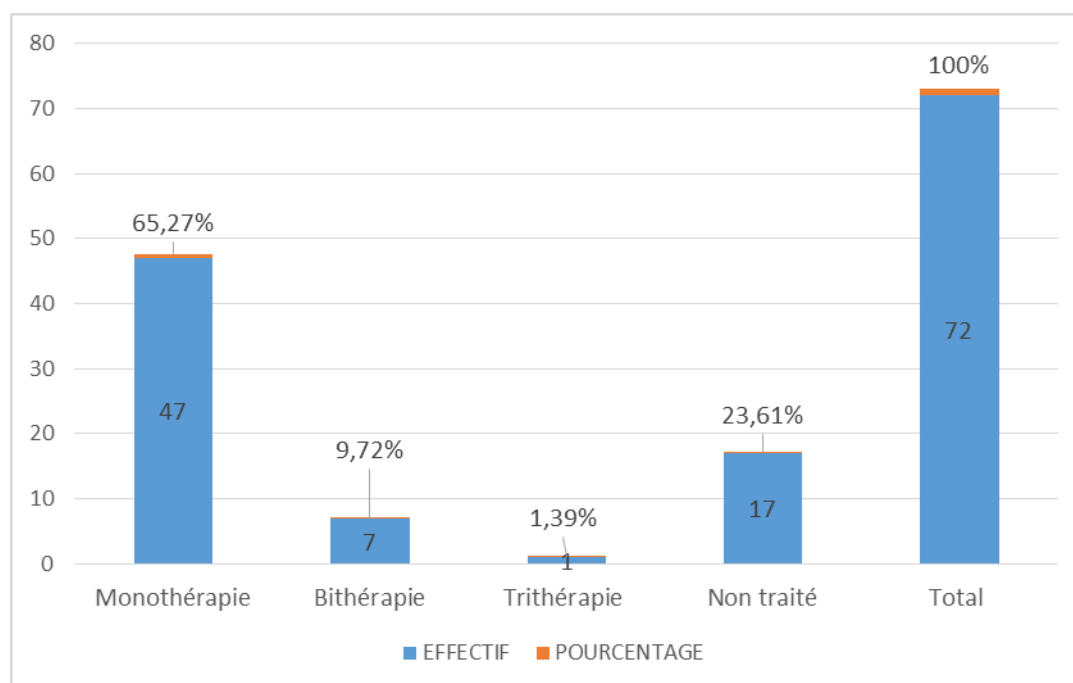
This Fig. 4, originally a bar chart, would display the percentage of patients who followed different therapeutic paths (medication, traditional treatment, prayer, etc.).



**Figure 4:** Distribution of patients according to their therapeutic itineraries.

It appears from Fig. 5 that clinicians primarily resorted to monotherapy (65.27%) for the induction of epilepsy treatment, while the use of bitherapy and tritherapy represented only 11% in total, 9.72% in the first case and 1.39% in the second. Moreover, for certain reasons, patients had not received treatment (23.6%).

*c. According to therapeutic protocols*



**Figure 5:** Distribution of patients according to the therapeutic protocol used.

*d. According to duration of consultations*

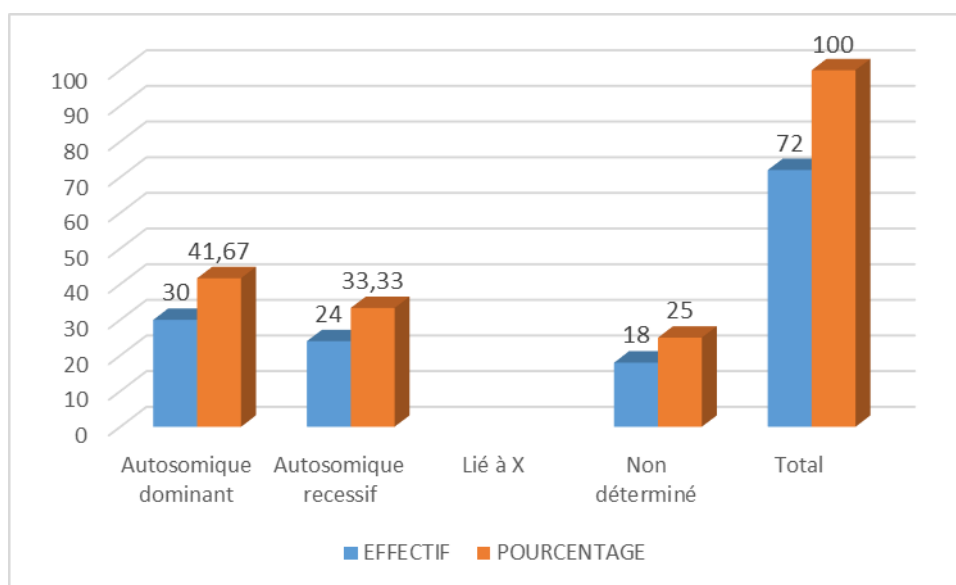
The following table shows the results between the onset of epilepsy and the date of consultation. We observed that in 73.61% of cases, patients and/or families of patients living with epilepsy were only able to come to the hospital more than one year after the onset of the disease. The remaining 26.4% represented durations of less than 6 months and between 7 and twelve months, respectively.

Duration between onset of disease and date of consultation	n	%
Less than 6 months	9	12,50
Between 7-12 months	10	13,89
Greater than 12 months	53	73,61
Total	72	100

**Table 3:** Distribution of patients according to the duration of consultations.

*e. According to mode of transmission*

In Fig. 6, we considered the autosomal dominant and autosomal recessive modes. We deduce from this Fig. 6 that epilepsy is transmitted in the autosomal dominant mode in 41.67% of cases and in the autosomal recessive mode in 33.33% of cases.

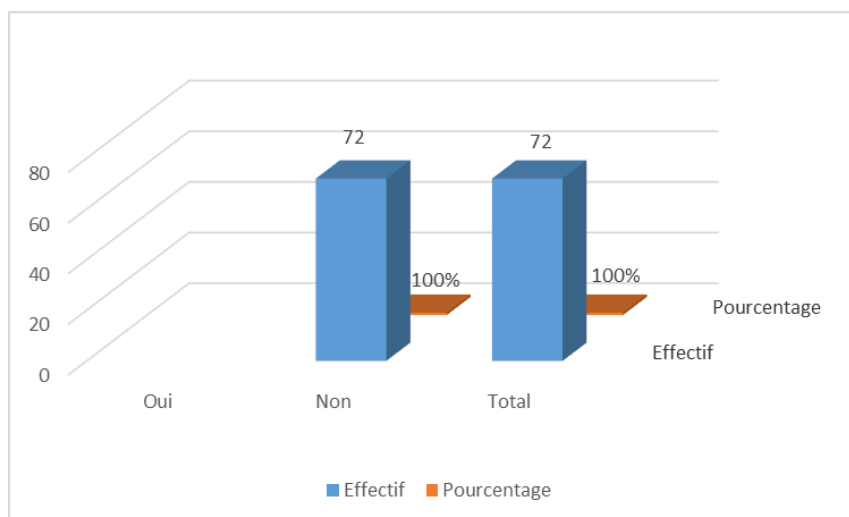


**Figure 6:** Distribution of patients according to the mode of transmission by pedigree.

*f. According to consanguinity between parents*

The results regarding the notion of consanguinity between the parents of patients living with epilepsy are shown in Fig. 7, indicating that no patient resulted from a consanguineous marriage.





**Figure 7:** Distribution of patients according to the notion of consanguinity between the parents of patients living with epilepsy.

Distribution of patients according to two associated parameters (Table 4).

Types of Seizures																	
	ABSENCE		CGT C		ATONIC		P. C		P.S. G		P. S		TONI C		MYOCLONI C		TO T
	n	%	n	%	n	%	n	%	n	%	n	%	n	%	n	%	72
F	2	6,4 5	20	64,5	2	6,45	1	3,2 3	2	6,4 5	2	6,4 5	1	3,2 3	1	3,23	
M	4	9,7 6	31	75,6	1	2,44	2	4,8 8	1	2,4 4					2	4,88	
	6		51		3		3		3		2		1		3		
CGTC: Generalized Tonic-Clonic Seizures, PC: Complex Partial Seizure, PS: Simple Partial Seizures, F: Female, M: Male.																	

CGTC: Generalized Tonic-Clonic Seizures, PC: Complex Partial Seizure, PS: Simple Partial Seizures, F: Female, M: Male.

**Table 4:** Distribution of patients according to sex and seizure type.

Reading this table shows us that generalized tonic-clonic seizures are present in considerable proportion in both sexes (64.5% of cases for females and 75.6% of cases for males) followed by absence seizures in males with 9.76% of cases, for a corrected chi-square of 6.1.

According to mode of transmission and seizure type (Table 5).

Types of Seizures																	
	ABSEN CE		CGTC		ATON IC		P.C		P.S. G		P.S		TONI C		MYOCLO NIC		TO T
	n	%	n	%	n	%	n	%	n	%	n	%	n	%	n	%	72
TD	2	6,4 5	21	70	2	6,4 5	2	6,4 5	2	6,4 5					1	3,3 3	
TR	3	12, 5	15	62, 5	1	4,1 7			1	4,1 7	2 8,33		1 4,17		1	4,1 7	
IN	1 5,56		15 83,3				1 5,56								1 5,56		
TO T	6		51		3		3		3		2		1		1		
TD: Dominant Transmission, TR: Recessive Transmission, ID: Undetermined																	

TD: Dominant Transmission, TR: Recessive Transmission, ID: Undetermined

**Table 5:** Distribution of patients according to mode of transmission and seizure type.

It appears from this table that CGTC are transmitted in 70% of cases according to the autosomal dominant mode and in 62.5% of cases according to the autosomal recessive mode, while absence seizures are transmitted in 12.5% of cases according to the autosomal recessive mode, with a corrected chi-square of 11.46.

## Discussion

### *Prevalence of Familial Epilepsies*

Our study collected 72 families with a history of familial epilepsy, out of a total of 641 patients diagnosed with epilepsy during consultations, representing a prevalence of 11.23% of cases. This result is consistent with other authors, notably M Ndiaye, et al., in Senegal, who found a general frequency of familial epilepsies of 11% in their study. The same observation was made by Kuate-Tegueu Callixte in 2014 in Cameroon in his study on epilepsy, who found that more than half (57.1% of cases) of patients living with epilepsy had a family history of epilepsy and among these, 11.4% of cases had two family members suffering from epilepsy and in 10% of cases, some had three or more patients suffering from the same epileptic disease [39,40].

**Notion of consanguinity** While Lamine Thiam in Senegal in 2020, in his study on the clinical and paraclinical aspects of idiopathic epilepsy in children at the Hôpital de la Paix in Ziguinchor, showed that 18 children were followed for idiopathic epilepsy and that parental consanguinity and family history of epilepsy were found in 1 and 6 children respectively (i.e., 5.5% and 33.3% of cases). This leads us to say that the prevalence of 11.23% of cases in our environment without a notion of consanguinity, as shown in Fig. 7, between parents in our environment, pushes us to seek a hereditary character that needs to be demonstrated and to identify the genes and/or variants involved. This is why, for several years, the role of ion channels in epilepsy has been suspected, mainly on physiological arguments, because ion channels (Na<sup>+</sup>, K<sup>+</sup>) are responsible for the generation of action potentials, neurotransmitter receptors are themselves channels or are coupled to ion channels and many antiepileptic molecules act directly or indirectly on their functioning. Recent molecular genetics data have confirmed this role. Indeed, the study of familial forms of epilepsy has made it possible to incriminate genes that code for subunits of ion channels or subunits of channel receptors [41,42].

In their studies on epilepsy, a family history of epilepsy was found 2 to 3 times more often in patients with epilepsy than in controls [43,44]. It has been shown in the literature that consanguineous marriages are common in certain cultures in Asia, particularly in Indian or Muslim communities. In a study in India, consanguineous parents were more frequent among patients than among controls (13.1% vs. 6.6%) (2.6; 95% CI: 1.5-4.4) [45]. The association with consanguinity is stronger in generalized epilepsies than in localized epilepsies and in idiopathic or cryptogenic epilepsies than in symptomatic epilepsies [46,47].

Average age and sex as shown in Table 1 and Fig. 2, in our cohort, the average age at diagnosis of patients living with epilepsy is 15.24pm 11.03 years. With a maximum of 75% at 20 years and a mode of 17 years. For her part, Zubcevic, 2009, reported an average age at diagnosis of 5 years. Many other studies report a high frequency of hereditary seizures in children [48].

In their studies, Dadah Samy Mohamed Lemine, et al., and Mukuku O, et al., reported an average age of patients living with epilepsy in Senegal that ranged around 8 years, with a peak found in the 5 to 10 year age group. Also, male predominance as we demonstrated in our study with a sex ratio of 1.32 [48-50]. This male predominance found is explained by the possible neurobiological difference between the neurons of male and female subjects leading to a differentiation of responses during brain lesions [51]. This male overrepresentation could also be explained in our environments by an underreporting of the disease in young women of marriageable age. For their part, Amina Chentouf, et al., in Algeria found in their study on the clinical characteristics and hereditary profiles of epilepsies a sex-ratio of 1.35 in favor of the male sex without a notion of X-linked transmission [52].

**Therapeutic itinerary, drug treatment, time between onset of disease and day of consultation.** The results recorded in Fig. 4 of our study showed that in 48.61% of cases, patients followed a drug treatment before arriving at the hospital and that traditional treatment and prayer accounted for a significant proportion, namely 36.11% and 9.72% of cases. Furthermore, in the quest for healing, patients find themselves in conditions where they combine several treatment methods. The cumulative percentage of patients who followed the path of prayer and traditional treatment sufficiently shows the significant proportion of presenting for consultation beyond 12 months, as shown in Table 5 of this study.

Moreover, this disease is associated with several false beliefs that lead to stigmatization that the evolution of scientific knowledge cannot change in Africa. Thus, we believe that many people with epilepsy live hidden in the shadows due to the social burden, which greatly impacts many studies because only a minority will come to the hospital for treatment. Furthermore, it is known that beliefs, perceptions and apprehensions vary from one country to another and can in all cases influence individual and collective strategies regarding treatment options [53].

At the medical level, the treatment of epilepsy faces various obstacles, whether it is the cultural perception of the disease, the low priority given to it by health authorities, infrastructure deficiencies or the irregularity of drug supply. Therefore, we believe that the high cost of medicines, the lack of social coverage, distance, the unequal distribution of health structures, all these situations associated with the lack of qualified personnel make the situation of people with epilepsy difficult, as other authors have pointed out [48].

Epilepsy, as a chronic disease that requires long-term care, seems unusual in Africa or even poorly accepted and therefore, the social impact of the disease in Africa, the social and cultural consequences of the disease are extremely detrimental to epileptic patients. They can be broadly summarized in one word: rejection or stigma. Rejection also comes from the family circle, although patients are almost never driven out of their homes and rarely sleep in a separate place ("disease from behind the house"). It is not real exclusion but rather marginalization. The patient is never banned, regardless of the society studied. They are tolerated, but no longer have a role in society (Andriantsimahavandy A, 1997). Patients suffer from this marginalization. They are ashamed of their illness, often feel diminished and therefore hide the diagnosis from others. The epileptic in Africa is a deviant whose existence is tolerated. They are a model of collective anxiety and social surveillance. Because of its frequency and heavy socio-economic consequences, epilepsy in developing countries like the Democratic Republic of Congo still represents a major public health problem [47]. Therefore, a community approach and widespread public awareness will be needed to effectively manage the problem of epilepsy. From the analysis of Fig. 5, it appears that clinicians primarily resorted to monotherapy (65.27%) in the induction of epilepsy treatment, while the use of bitherapy and tritherapy represented only 11% in total, 9.72% in the first case and 1.39% in the second. The induction of anticonvulsant treatment by monotherapy was also found by other authors, notably M Ndiaye, et al., and M Bourrous, et al., who, in their respective studies, found that monotherapy was the most used therapeutic regimen and that valproic acid was the most commonly used molecule [44,45].

The data from our study show that a significant proportion of patients living with epilepsy, namely 23.61% of cases, did not receive treatment, certainly for multiple reasons that could also find some explanations as mentioned in Fig. 4 and therefore, the reasons for this therapeutic insufficiency in sub-Saharan Africa seem complex and multifactorial. They involve socio-cultural, economic causes and even the inaccessibility and inadequacy of health structures. It is therefore important, in our opinion, to continue research in this area as part of the fight against epilepsy. As early as 1994, Danesi MA, et al., in their survey on epilepsy and population attitudes, showed that in 57% of cases, patients admitted to first resorting to traditional medicine. Moreover, to justify this attitude, in addition to beliefs, 43% of those surveyed cited the high cost of anti-epileptic drugs. Indeed, all socio-cultural factors and especially the supposed (supernatural) causes of epilepsy, can explain the recourse to traditional therapists and/or the combination of several treatment modalities in the quest for a solution. Dale JR, on the other hand, in 1984, in his study conducted among Tanzanian traditional practitioners, showed that they often took care of patients living with epilepsy, while epilepsy was not listed as a health problem in the modern health care system. Therefore, it is normal to understand here the difficulty of conducting a good study on the prevalence and/or incidence of epilepsy in many areas, because generally, it is the first recourse (traditional medicine and/or prayer) and only the observation of several failures could motivate a hospital consultation.

### *Type of Seizures*

We found that generalized tonic-clonic seizures were the most common with 70.83% of cases, followed by absence seizures with 8.33% of cases. The frequency of other seizures ranged between 1.39% and 4.17% of cases. In his study on epilepsy and schooling in Congolese epileptic children, Ntenga Parice found that generalized tonic-clonic seizures were the most common with 41.7% and a p-value of 0.04, followed by absence seizures with 33.3% and a p-value of 0.67. This strong representativeness of generalized tonic-clonic seizures would be justified by their spectacular nature and thus easy recognizability, but also by the population's lack of knowledge of other types of seizures. This same observation has been made in most studies conducted in Africa (Dadah Samy Mohamed Lemine (Senegal, 2015), Chentouf A [39].

### *Mode of Transmission of the Pathology According to the Pedigree*

We noticed from Fig. 6 that the autosomal dominant mode of transmission represents 41.67% of cases and that transmission according to the autosomal recessive mode is around 33.33% of cases. This result contradicts that of MEFOUNG Ephrata Samuel, 2021, for whom the autosomal recessive mode of transmission represented more than half of the cases. On the other hand, our results are comparable to those of a study conducted in Algeria by Chentouf A, et al., who found a high prevalence in favor of the autosomal dominant mode. And so, this leads us to say that these differences could be justified by the sample sizes which are different to characterize a national distribution and highlight the predominance of one mode of transmission over the other [40].

### **Conclusion**

We conducted a prospective, cross-sectional analytical study from October 2020 to October 2024, which included epilepsy cases presenting for consultation during the study period. Patients aged 40 years or younger with epilepsy and their families who were seen at the aforementioned investigation sites, with a notion of familial epilepsy discovered or reported during the consultation, were considered as potential candidates for this study. We performed a stratified probabilistic sampling during this study to select the subjects who participated. A total of 72 families were collected and subjected to statistical analyses. This allowed us to demonstrate that:

- The prevalence of familial epilepsy in Lubumbashi is 11.23% of cases.
- The mean age of patients with epilepsy and a family history of the disease is  $15.24 \pm 11.03$  years, with a median of 14 years, a 1st quartile of 7 years and a 3rd quartile of 20 years.
- The sex ratio is 1.32, favoring males.
- Generalized tonic-clonic seizures came in first place, with 70.83% of cases, followed by absence seizures with 8.33% of cases. The frequency of other seizure types ranged from 1.39% to 4.17% of cases.
- The EEG was performed in the majority of patients (63.49% of cases) and was normal in 27.78% of cases and abnormal (presence of paroxysmal grapho-elements) in 35.71% of cases. The exam was not performed in 38.57%.
- In 48.61% of cases, patients were on medication before arriving at the hospital and traditional treatment (36.11%) and prayer (9.72%) accounted for a total proportion of 45.83%.
- Clinicians primarily resort to monotherapy (65.27%) to initiate treatment for epilepsy, while the use of dual and triple therapy accounted for only 9.72% and 1.39% of the total, respectively. Furthermore, for various reasons, some patients had not received any treatment (23.6%).
- In 73.61% of cases, patients and/or their families were only able to come to the hospital more than one year after the onset of the disease. The remaining 26.4% represented a duration of less than 6 months and a duration ranging between 7 and 12 months.
- Epilepsy is transmitted in an autosomal dominant and recessive manner in 41.67% and 33.33% of cases, respectively.
- No patient came from a consanguineous marriage.

### **Perspectives**

The analysis of this epidemio-clinical data on familial epilepsy in Lubumbashi is essential and impacts understanding. These data should be the subject of further investigation with a genetic study to understand the specifics.

### **Conflict of Interest**

The investigators declare no material or financial conflict of interest related to this study.

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