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Neuro-Imaging of Phacomatoses: about 20 Cases

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Summary

Phacomatoses are a group of congenital diseases that are very different genetically, clinical-ly and radiologically. They are responsible for the formation of hamartomatous or tumor lesions, the preferential impairment of which is the central nervous system and the skin. Neurological manifestations are often a major component of the prognosis. Imaging, including MRI, allows diagnosis, patient monitoring and screening in family members. The most common diseases reported in our article are neurofibromatosis type 1 and 2, Bournevilletubepink sclé,von Hippel-Lindau disease and Sturge-Weber syndrome.

Keywords: Phacomatoses; Imaging; MRI; Tumor

Work Objectives

To write the different radiological aspects on the brain and spinal floor of the main phacomatoses:

Neurofibromatosis TYPE I

Neurofibromatosis TYPE II

Bourneville Tuber Sclerose

Sturg Weber's Disease

From Hippel Lindau's Disease

Define the place of imaging in the diagnosis and management of patients with the disease

Materials and Methods

This is a descriptive respective study conducted over a 6-year period (2014-2019) at the Medi-Cal Imaging Department at the

Mohammed VI University Hospital in Oujda, Morocco, involving 20 cases of phacomatoses with recognized diagnostic criteria: Neurofibromatosis type 1 (10cas-es); cas); Neurofibromatosis type 2 (1cas); Sclérose tub Bourneville tuberous pink Scl (7cas-es); Sturge weber disease (1 case); Von Hippel Lindau disease (1 case).

(TDM) cere19 patients were explored by magnetic resonance imaging (MRI) on a GE signa HDXT 1.5 Telsa 16-channel device and 1 patient received a CT scan. brale.

Results

Age

The age of patients in our series varies between 2 years and 49 years (with an av-erage age of 25.5 years), the majority of patients with phacomatosis had an age between 0 and 10 years or 65%(Figure1).

Sex

In our series, there is a slight predominance of the male sex; 10♂/9♀ with a sex ratio of 1.1 (Figure 2).

Reason for consultation

In our series we note the following distribution according to the reason for consultation: Seizures (no.10) or 53%; a chromic spots (no.3) or 16%; Delay in psychomotor acquisi-tions (no.5) or 26%; Facial swelling (no.2) or 11%; Polyglobulia (No.1) or 5%; Angioma plan cutanfacial (no.1) or 5%; decrease in visual acuity (no.1) or 5%; headache and balance disorder (no.1) or 5%.

Imaging Data

Observation 1: Neurofibromatosis type 1

a 4-year-old child is admitted for delaying psychomotor acquisitions, arable cereatic MRI has been performed (Figure 3).

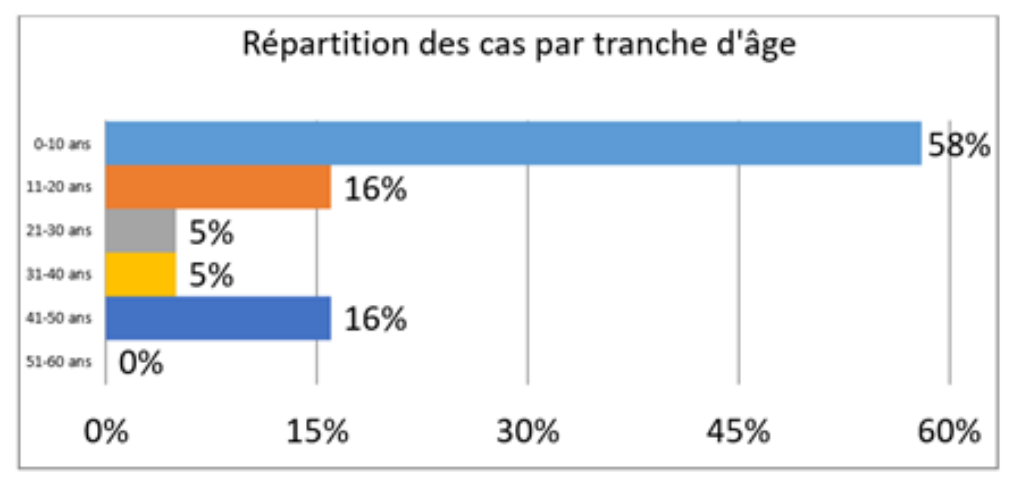

Figure 1: Breakdown of cases by age group.

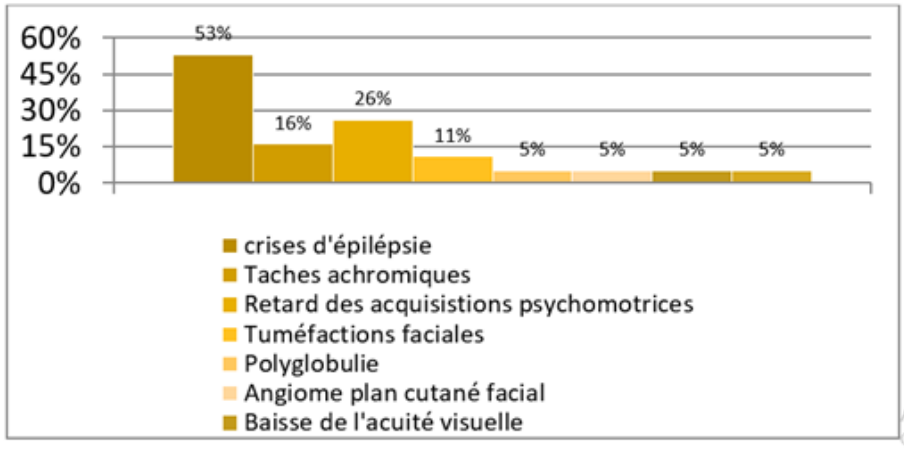

Figure 2: Distribution of patients based on the reason for consultation.

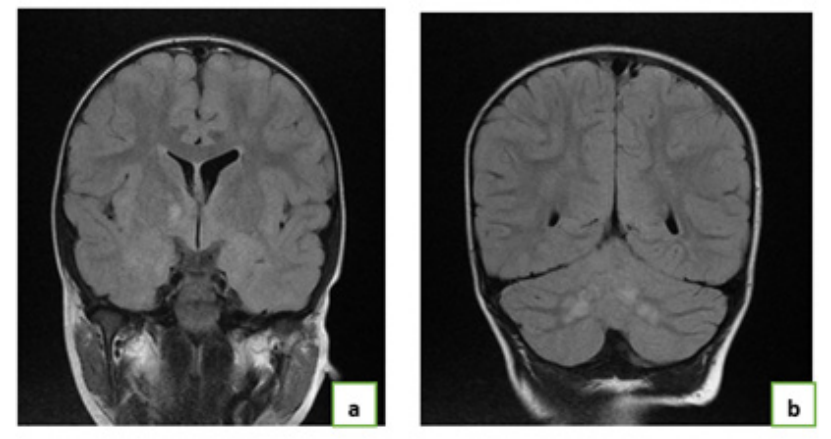

Figure 3: T2 weighted axial MRI of the brain T2 (a) and FLAIR (b, c) showing a triventricular hydrocephalus with transependymal resorption associated with signal abnormalities in T2 and FLAIR hypersignal of the internal capsules predominantly bilaterally on the side in relation to OBNI

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Observation°2: Neurofibromatosis type 1

A 10-year-old child who senses mental retardation associated with a chromic stain and epi-leptic seizures. The a Bennyficie of an MRI CéréDude (Figure 4).

Observation°3: Neurofibromatosis type 1

A 7-year-old child is admitted for seizures, followed by psychomotor delay, clinical examination reveals the presence of a chromic spots. Hereceiveda cerebraleMRI (Figure 5).

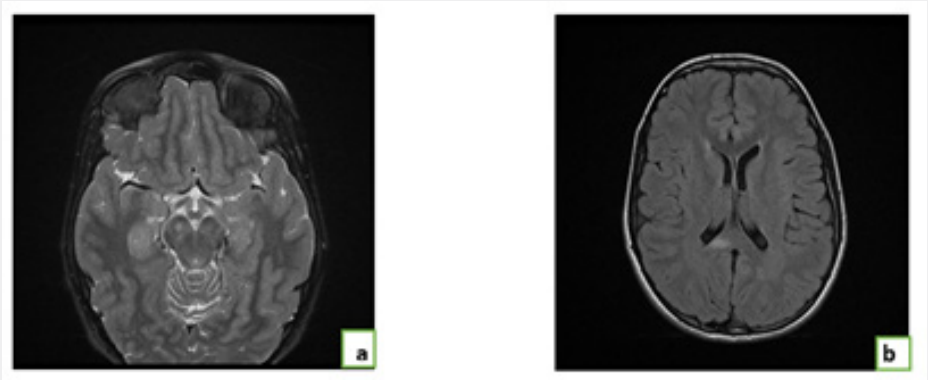

Figure 4: T2(a)and FLAIR(b)axial cup MRI showing signal abnormalities of the right cerebral peduncle, callous body spleen and left lenticular nucleus in hypersignal T2 and FLAIR in relation to "OBNI".

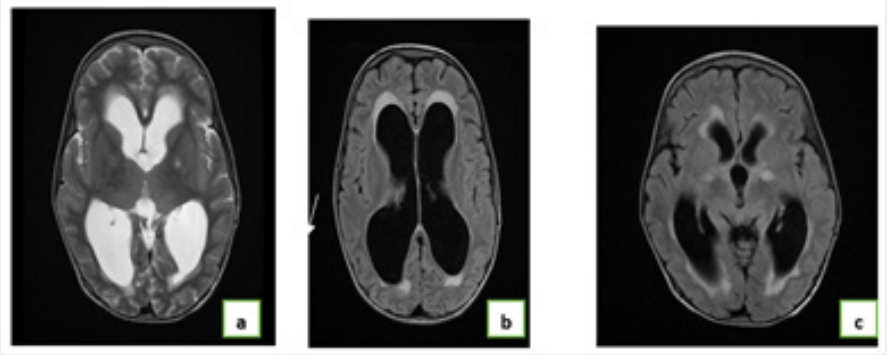

Figure 5: MRI of the axial cup T2(a)and FLAIR(b, c)showing tri-ventricular hydrocephalus ave-cresorption trans-ependymal) et FLAIR (ass coupled withée s X2 hypersignal signal abnor-malities and FLAIR of internal bilaté-perminating bilaee on rale the left side in relation to "OBNI".

Observation°4: Neurofibromatosis type 1

A 7-year-old girl is admitted for a delay of psychomotor acquisitions, anencephalic I RM has been performed (Figure 6).

Observation°5: Neurofibromatosis type 1

A Child Aged 9-year-old follow-up for neurofibromatosis type I and which has a left cervical swelling in rétroauriculaire soft consistency associated with stains coffee at Milk. Udoes MRI created Dude was achieved (Figure 7).

Observation°6: Neurofibromatosis type 1

patient 9 years of age followed for neurofibromatosis type 1 with epileptic seizures associat-ed with achromic spots. A Brain MRI with injection of contrast product it has been carried out (Figure 8).

Observation°7: Neurofibromatosis type 1

P49-year-old Years follow-up for neurofibromatosis type 1. A Brain MRI was achieved in the

Patient (Figure 9).

Observation°8: Neurofibromatosis type 1

An elderly patient 12-year-old follow-up for neurofibromatosis type 1 admitted for de-creased acuity visual left. A Orbital MRI was performed (Figure 10).

Observation°9: Neurofibromatosis type 1

A e 19-year-old girl who was being followed for NF1 and who had been exhibiting bilateral lumbosciatic pain low back pain for the past bilatéyear, accentuated by walking. rale accentu A spinal mri was per-formed (Figure 11).

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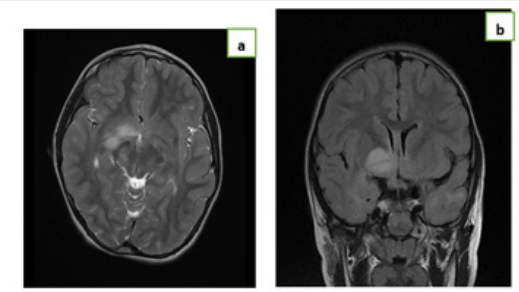

Figure 6: Brain MRI T2 in axial T2(a)and coronal SIGNAL(b)brain MRIshowing multiple bilateral diffuse signal ab-normalities in T2 and Flair, in unenhanced T1 hypo signal after Gadolinium injection in rela-tion to brain hamartomes returning toNF1.

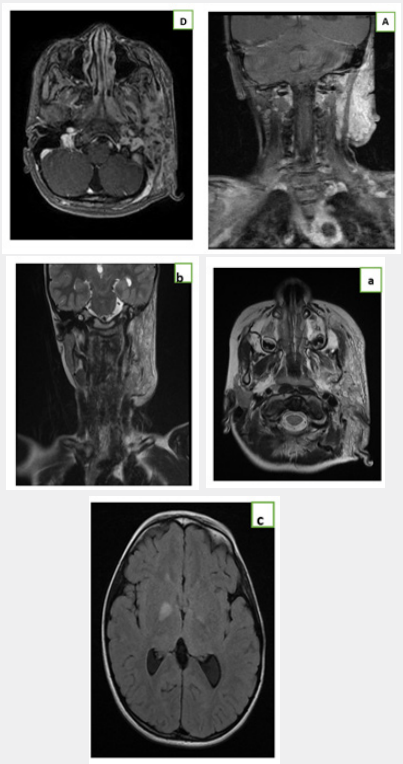

Figure 7 : IRM cerebral T2 axial and coronal cut (a and b), axial FLAIR (c); showing a volumi-nous mass latero-left cervical infiltrates the homolateral hemiface and extends to the deep spaces of the face in relation to a neurofibroma associated with abnormalities of the white substance in relation to OBNI.

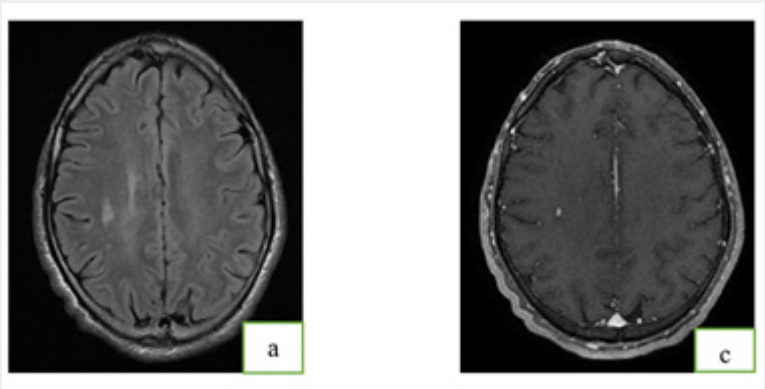

Figure 8: Brain MRI in axial cut T2 FLAIR (a) objectifying signal abnormalities of the SB on the above tensorial floor in relation to OBNI with detection of micronodular contrast after injection of Gado (b).

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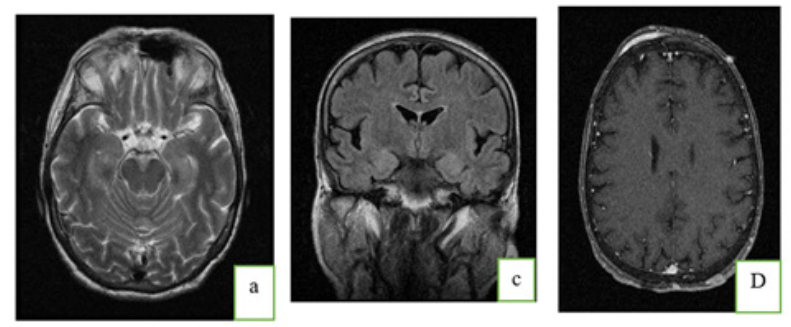

Figure 9: Brain MRI in Axial Cut T2 (a), coronary FLAIR (b) and injected axial (c) showing the presence of multiple nodular lesions under the skin in relation to neurofibroma associated with hippocampal hypertrophy as part of its pathology

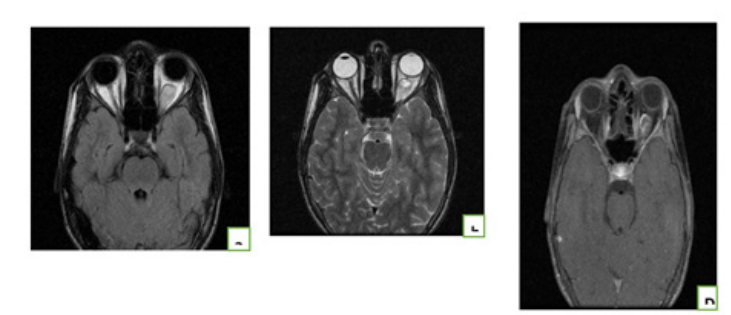

Figure 10: cerebral MRI in axial cut FLAIR (a), axial T2 (b), coronal T2 (c) and injected axial (b) showing an intra-conical, intra-orbital tissue process centered on the optic nerve that enhances in a heterogeneous manner in relation to an optic nerve glioma.

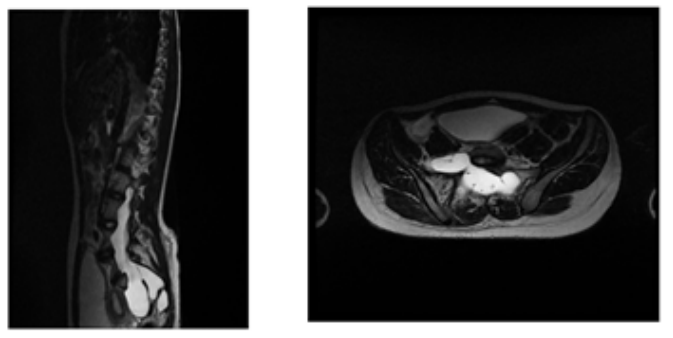

Figure 11: Medullary MRI in sagittal cut T1 (a), sagittal T2 (b), axial T2 (c) showing a dural ectasia lombosacreted.

Observation 10: Neurofibromatosis type 1:

Patient of 26 followed for a functional impotence of the right upper limb, with a reduction of the progressive installation walking perimeter, without notion of sphincteric disorders (Figure 12).

Observation11: 1 Neurofibromatosis type 2:

Patient aged 46 years, has BAV, right ptosis, bilateral tinnitus and deafness as well as low back pain. Acerebro-spinal MRI was performed (Figure 14).

Observation No. 12: Bourneville-Pringle diseasel

A 6-year-old child followed for repeated epileptic seizures, and as part of an etiologi-calcheck-uphe received an encephalic MRI;

Observation No. 13: Bourneville-Pringle disease

A2-year-old girl is admitted for psychomotor delay and seizures with diffuse achromic spots. Theepileastial ENc MRIinsagittal cut T1, axial T2 and diffusion, coronal T2 FLAIR showed: (Figure 5) (Figure 15)

Observation No. 14: Bourneville-Pringle disease

A 4-year-old girl is admitted for epileptic seizures without a found cause, a T1 sagittal cut brain MRI, axial T2 and diffusion, coronal T2 FLAIR was done Figure 16.

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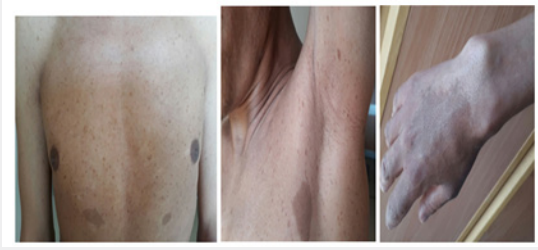
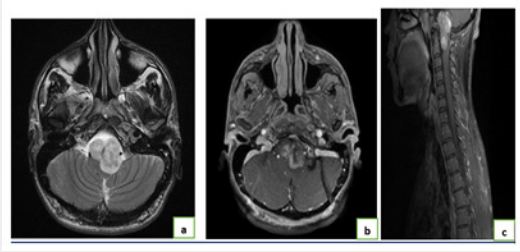


Multiple coffee-to-milk spots all over the body, axillary lentigines and sub-decious nodular neuro-fibromas.



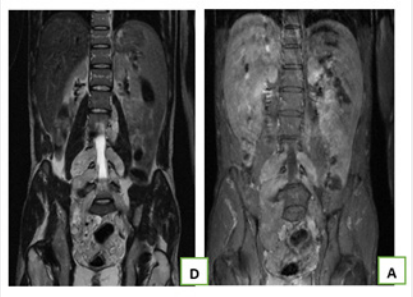
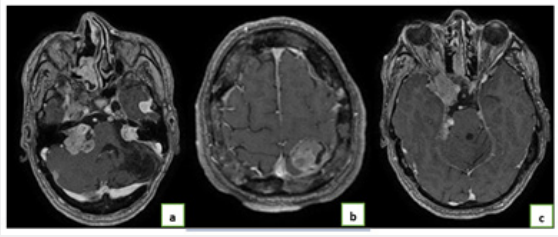


Figure 12: Brain MRI cut axial T2 (a) and injected (b), injected sagittal cut (c): neurofibroma in hypo signal T1, hypersignal T2, enhancing in a heterogeneous manner after injection of contrast product, extended through the C1-C2 conjugation foramen compressing the bulb and the cervical bone marrow. T2 (d) and injected corneal medullary MRI (e): Multiple extended Storied neurofibromas from D12 to the bilateral sacred e.



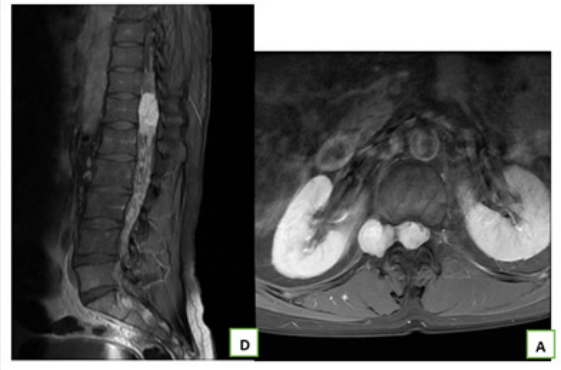


Figure 13: Injected axial cut brain MRI (a, b and c) showing multiple over speed and tentorial processes associated with bilateral vestibular scwannomes. Sagittal (D) and axial (e) medullary mri scan showing neuromas, meningeal thickening and epondymomas.

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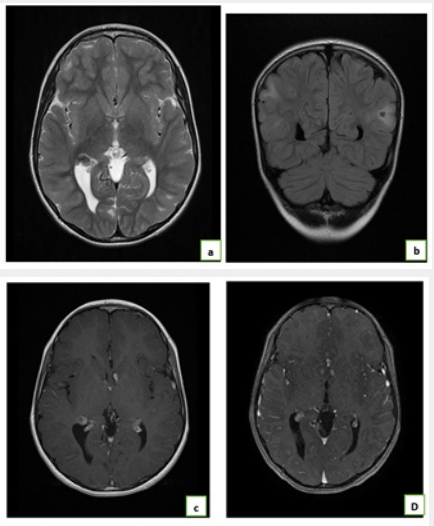

Figure 14: Brain MRI showing multiple nodules under heterogenic T2 signal ependymal(a)are enhanced after injection of gadolinium (cc ne T2 (and d)associated with cortical signal ab-normalities in ranges, triangular in hypo signal T1, hypersignal T2 and FLAIR (bb),not enhanced after injection of gadolinium, in relation to cortical tubers re-entering the context of a STB.

Observation No. 15: Bourneville-Pringle disease

A 14-year-old child followed for seizures, a brain MRI was performed (Figure 17)

Observation No. 16: Bourneville-Pringle disease

A 35-year-old woman followed for STB, who has bilateral frontotemporal facial swell-ings with right exophthalmia. A brain scan in axial cuts of 1 to 3 mmthick without and with injection of PDC with multi-planar reconstructions was carried out objectively: (Figure 8) (Figure 18)

Observation No. 17: Bourneville-Pringle disease

E nfant aged 9 years, followed for epileptic seizures. a brain MRI was performed (Figure 19) and hide the patient's eyes.

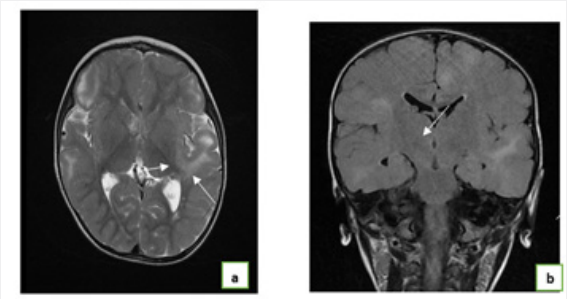

Figure15: Brain MRI in Axial Cut T2 (a),coronal FLAIR(b):nodular formations under ependymals protruding into ventricular light, T2(a),enhances after injection (associated with ne T2 (cortical signal abnormalities in ranges, triangular in hypersignal T2 and FLAIR(b),not enhanced after gadolinium injection, in relation to cortical tubers.

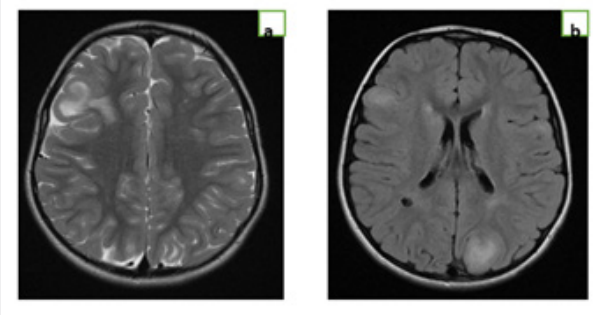

Figure 16: Axial cut MRIT2(a), FLAIR(b)showing multiple cortical ranges of the white substance sus ten-torial in), FLAIR (hypo signal T1, hypersignal T2(a)and FLAIR (bb) without restriction of diffusion and without taking pathological contrast correspondent to corsical tubers.

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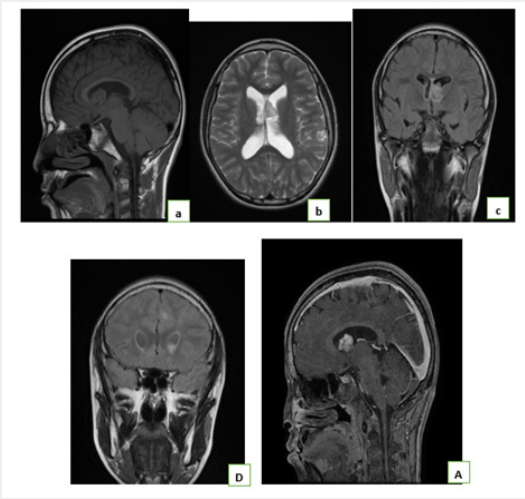

Figure 17 :T1 sagittal cut ceralMRI before(a)and after injection of gadolinium (e),axial T2(b)and FLAIR coronals (cc and d)showing a giant cell astrocytoma of the left lateral ventricle above Monro's hole, in hypo signal T1 (a)and heterogeneous signalT2(b) and-FLAIR (cc), enhanced after injection of gadolinium (ee),associated with signal abnormal-ties of the cerebral cortex visible only on the sequence FLAIR(d) in relation to cortical tubers.

Observation No. 18: Bourneville-Pringle disease

26-year-old patient, followed by epileptic seizures and e de 26 (Figure 20)

Observation n° 19: Syndrome de Sturge Weber

A three-year-old girl with no particular pathological history is admitted for simple partial seizures. Clinical examination reveals the presence of a cut a facial plan angioma median extending over the upper eyelids. A brain CT scan without injection of the contrast product is performed (Figure 9) :) (Figure 21). CT scans show subcortical calcifications off rondo-temporo-parieto-occipital gyri form appearance, bilateral bilat, bilateral enlargement of subarachnoid peri cerebral spaces, in-creased size of lateral ventricles, and bilateral hypertrophy of the choroid plexus. Faced with these CT scans and the presence of cutan angioma in V1 territory, the diagnosis of Weber Sturg-Weber syndrome / Von Hippel-Lindau Disease. was made.

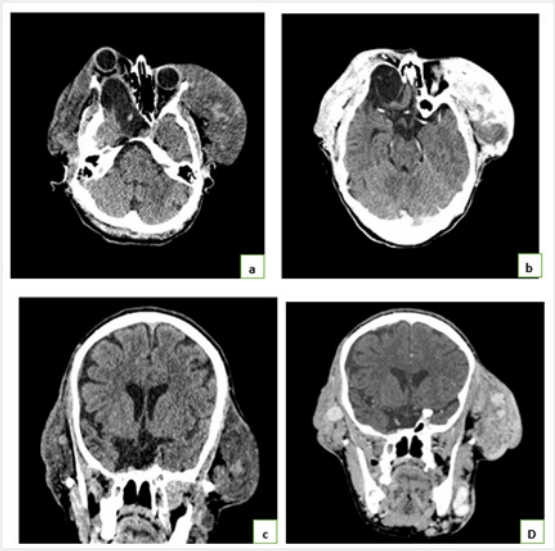

Figure 18: Cerebral CT scan in axial cut before(a)and after injection of PDC (b)and coronary before(c)and after injection of PDC (dd) showing bilateral frontal-temporal facial angio-fibroids heterogeneous hypodense(a and c),having intense contrast after PDC (c and d) associated with an unenhanced right frontotemporal cystic tuber after PDC exerta mass effect on the homolateral eyeball responsible for grade III exophthalmia (a and b).).

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Observation No. 20: Von Hippel Lindau Disease

A 48-year-old hypertensive diabetic man being followed for polyglobulia, he received brain scan. cérébral. The patient had also been given a thoracoabdomino-best scan (Figures 22,23).

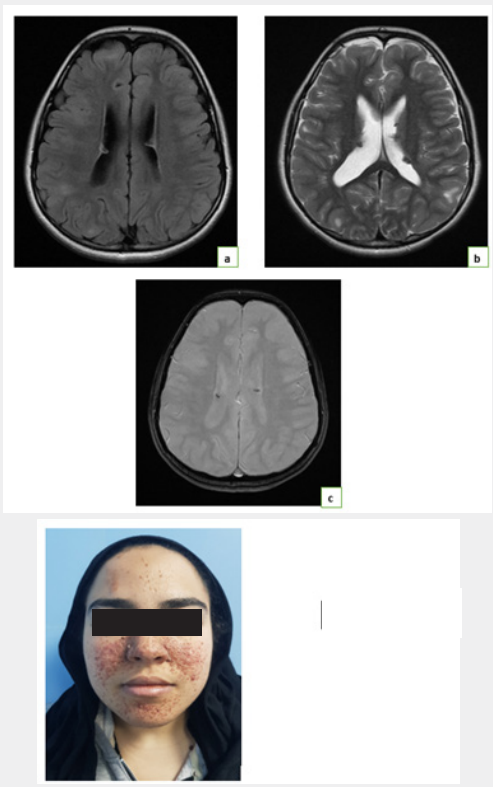

Figure 19: MRI of the Axial Cut FLAIR before(a), axial T2(b)and axial T2 (c)c objecting to signal abnormalities of SB on the tentoriel floor in relation to OBNI. cortical tubers and nodules under partially calcified ependymal. 26-year-old patient followed by epileptic seizures and e de 26.

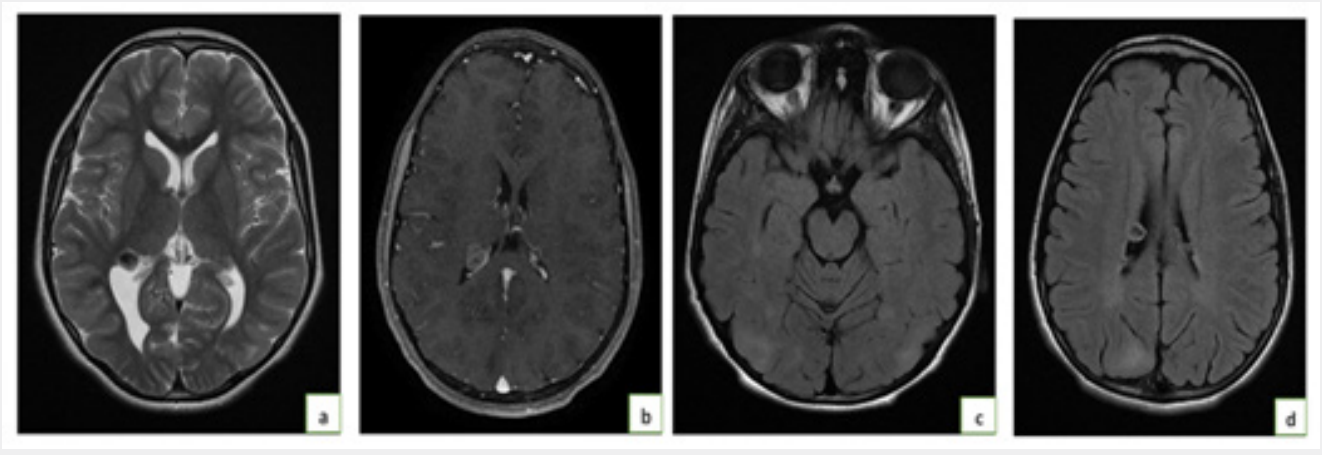

Figure 20: MRI of the axial cup T2(a)T1 after gadolinium injection(b)showing a giant cell astrocytoma of the right lateral ventricle. axial cut FLAIR (c, d) showing signal abnormalities in relation to cortical tubers.

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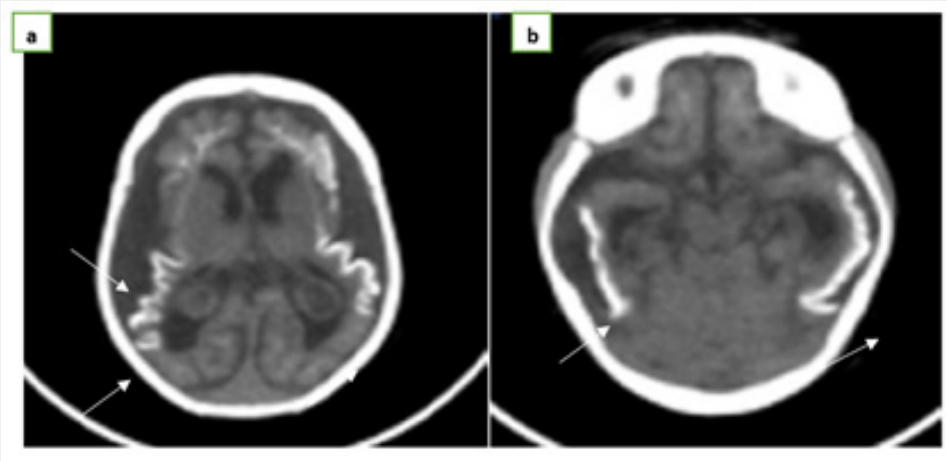

Figure 21: Brain CT scan without CDP injection shows subcortical calcifications off onto-temporo-parieto-occipital gyri form appearance, bilateral bilat, bilateral enlargement of subarachnoid pericrebrantal spaces, increased size of lateral ventricles and bilateral hypertrophy of the choroid plexus as part of Sturge Weber Syndrome.

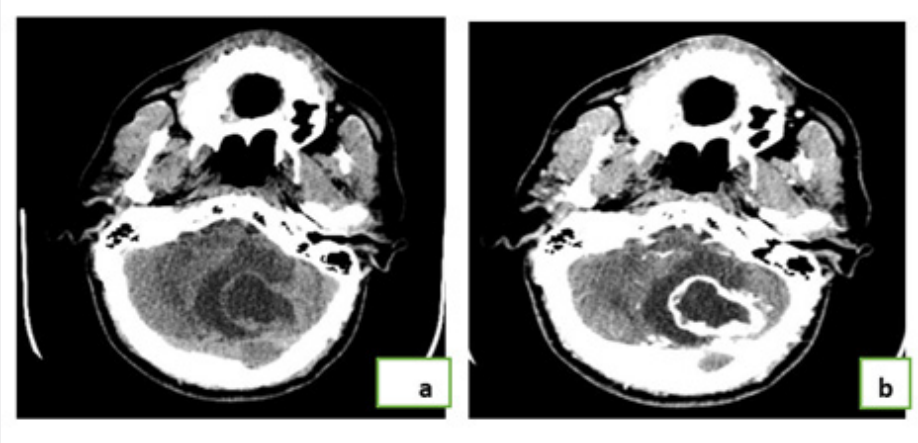

Figure 22: Cerebral CT scan before injection (a) and apéro injection of PDC (b, c) objectifying a volu-minous tumor process with a dual fluid component, which riphérique was modified after contrast and an ecrotic tissue component with central liquefaction and a fleshy peripheric tissue that enhances in intense-handled manner safter injection of PDC as part of von Hippel Lindau disease.

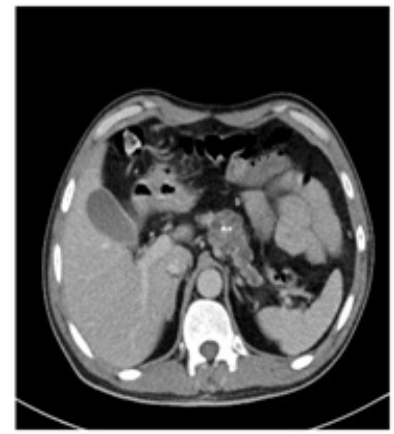

Figure 23: The patient had also been given a thoracoabdominal-best scan. Abdominal MT axial cut showing multiple microcysts of the pancreas performing an appearance in grape clusters with central calcifications in relation to a pancreatic serous cystadenome re-entering the context of its pha-comatous pathology.

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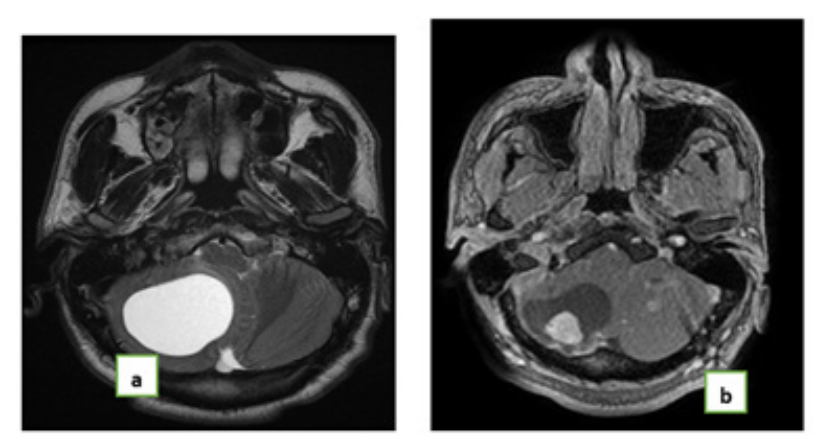

Figure 24: Cerebral MRI; sagittal cut T1 (a), axial T2 (b) and T1 after injection of Gado (c) showing voluminous solid-kystic cerebellar mass right with a wall nodal that rises intensely after injection in relation to a complicated cerebellous hemorrhic hegioblastoma of hydro-cephalus, which fits into the context of the disease of von Hippel. mangioblastome c.

Discussion

We will limit our analysis to the four most frequently encountered groups of conditions. Namely: neurofibromatosis type 1 and 2, Bournevilletubepink sclé,von Hippel-Lindau disease and Sturge-Weber syndrome.

Neurofibromatosis type 1 (NF1):

Neurofibromatosis type 1 (NF 1) or von Recklinghausen disease is the most common of genetic diseases, its frequency is about 1/3000 in the general population [1]. It is a dominant autosomal transmission genetic disease. The abnormal gene is located on the long arm of chromosome 17 (17q11.2). The protein coded by this gene, neurofibromin, intervenes in the control of cell differentiation and proliferation. The phenotypic expression is verysleea-ble [1,2]. It is characterized by a progressive and polymorphic impairment that can interest most organs with a predisposition for benign tumors and mylines [1]. The diagnosis of NF1 is essentially clinical and is based on well-defined criteria established at the 1987National Institute of Health Consensus Conference. Skin manifestations, particularly latte stains, epihbleds and skin neurofibromas, are at the forefront and often indicative of the disease. Neurological manifestations are also common, mainly represented by behavioral disorders, learning difficulties, seizures and symptoms related to tumor impairment of the optic pathways or even the hypothalamo-hypophysic axis [3,4]. Its management is multidisciplinary and imaging, and especially MRI, plays a prominent role in the initial assessment and monitoring of malignant-potential lesions [5,6].

NF1 Diagnostic Criteria

Despite the devability of the gene responsible for the disease, the diagnosis remains clinical and based on criteria defined by the 1987National Institute of Health (NIH)consensus.

The diagnosis of NF1 is based on the presence of at least two of the following criteria:

a. at least 6 latte stains larger than 5 mm in diameter in pre-pubescent individuals and more than 15 mm in pubescent individuals.

b. two or more neurofibromas of any type or plexiform neurofibroma.

c. axillary or inguinal ephelics;

d. a glioma of the optical pathways.

e. two or more Lisch nodules;

f. a characteristic bone lesion, such as sphenoidal dysplasia, thinning of the cortical long bones with or without pseud arthritis;

g. a first-degree relative with NF1 according to the previous criteria.

Neurofibromatosis type 2

Neurofibromatosis type 2 (NF2) is a genetic disease with dominant autosomal transmission characterized by the presence of bilateral vestibular schwannomes. The incidence of NF2 was estimated at 1 per 25,000 births in a study conducted in the north-west region of Eng-land with a population of 4.1 million. In the same study, the prevalence of NF2 was 1 in 100,000 inhabitants, and the annual incidence was 1 new case diagnosed in 1,312,000 inhab-itants per year [7]. The NF2 gene is the only known gene responsible for neurofibromatosis type 2. This gene encodes a protein, called merlin or schwannomine, inhibiting the growth of certain tumors and composed of 595amins.s. Clinical symptoms appear around the age of 20-30, later than NF1. Revealing symptoms are often due to the absence of acoustic neuromas with deafness and dizzi-ness. Root pain, due to one or more lumbar neuromas, is more rarely a mode of entry into the diagnosis of NF2.

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Two types of clinical forms have been described:

A severe form (Wishart type) characterized by an injury before the age of 25, the development of more than 3 tumors, repeated surgeries, and few survivors after the age of 40; a moderate form (Gardner type) characterized by a later onset age, fewer and slow-growing tumors, and many survivors after age 50. About 10% of NF2s are symptomatic before the age of 10. Skin signs are rare (about 30% of cases) compared toNF1, which is why the disease is often overlooked at first. Eye lesions (juvenile cataract) are frequent (50 to80% of cases). The diagnostic criteria for NF2 are strict, the main criterion being represented by a neuroma of bilatéral acoustics. However, this neurofibromatosis is characterized by multiple tumor damage to the central nervous system (meningiomas,e'pendymmes). Papillary pseudo-edema has also been reported to a gliotic pre-hair membrane [8].The following diagnostic criteria, called Manchester Criteras are necessary to establish the diagnosis early [8] bilateral vestibular schwannomes 2A first-degree parent with AND unilateral vestibular schwannome OR two ab-normalities: meningioma, schwannome, neurofibroma, glioma, cataract Unilateral vestibular schwannoma and two abnormalities: meningiome, schwannome, neurofibroma, glioma, cataract Multiples meningiomas AND unilateral vesti bular schwiomeOR two following abnormalities: schwannome, neurofibroma, glioma, cataract

Bourneville Tuberous Sclerosis (STB):

Bourneville tube-pink (STB) is the most frequent phacomatosisafterneurofibromatosis type 1, the incidence approaching 1/5800érose tub.

STB is a dominant hereditary disease of autosomal transmission, but 65-75% of cases are sporadic, corresponding to a mutation of novo [8]. There is genetic heterogeneity and genetic studies have currently identified 2 major genes, TSC1 and TSC2 [9,10].

This disease is characterized by dysplasias and/or neoplasias in the organs derived from the ectodermic leaf (skin, central nervous system Tre intandpe-ericic, eyes), structures derived from mesoderma (blood vessels, bones and cartilage) and endoderm (intestinal epithelium) may also be of interest. Central nervous system impairment is responsible for the most common clinical manifestations including epilepsy (West syndrome) and mental retardation.

Maladie de Sturge Weber (SSW)

Sturge-Weber syndrome (SSW), or angiomatosis encephalxeotrigor meningo-facial com-bines a plane angioma of a hemiface, in the territory of the tri-twin nerve (its upper branchV1), with a homolaterallepto-meningangioma and frequently a choroidal angioma. This condition is rare, its incidence is estimated at 1 /40,000. Pathogenesis is not known. This disease has no racial or ethnic predominance [11], it is con-genital, sporadic in most cases but family cases have been reported. It affects both sexes in the same way.

The angioma lepto-mening éing éingéing is constant, homolateral to the facial angioma, locatise at the level of the magpie mother most often at the occipital level but also at the parietal or even temporal lobe. The frontal lobe is usually spared. The cerebral cortex in front of it has a secondary atrophy that extends beyond the boundaries of the angioma, with ne-crosis and calcifications. Consequences of chronic ischemia, they are pericap and localized at the level of the 3rd and 4th atrophied cortical layers, and also at the level of the underly-ing white substance, neurons and vascular walls. There are outbreaks of cortical gliosis and white matter, is chic leons éand demyelination [12] (Table 1).

Table1: Diagnostic Criteria for Stb

Major Criteria	Minor Criteria
Angiofibromes of the face	Multiple geodes in dental enamel
Non-traumatic ungueal or peri-ungueal fibroids	Hamartomateous rectal polyps[3]
Hypo melanic stains (3)	Bone cysts[4]
Skin of sorrow	Radial migration lines in cerebral white matter [1.4]
Multiple retinal nodular hamartomes	Gingival fibroids
Tuber cortical [1]	Non-renal Hamartome [3]
Nodule sub-ependymaire	Achromic retinal stain
Giant cell astrocytoma	Skin lesions in "confetti"
Cardiac rhabdomyoma, single or multiple	Multiple kidney cysts [3]
Pulmonary Lymphangio-leiomyomatosis [2]	
Renal Angiomyolipoma [2]	

[1]When cortical dysplasia and white matter migration lines exist simultaneously, count only one criterion.re.

[2] When lymphangiomyomatosis and angiomyolipomes exist simultaneously, count only one criterion.re.

[3] Suggested histological confirmation.e.

[4] Sufficient radiological confirmation.

Maladie de Von Hippel Lindau (VHL):

Von Hippel Lindau disease (VHL) is a rare phacomatosis, it is a dominant autosomal genetic condition with variable expression, affecting1/36,000 to 1/40,000 people [13]. The Von Hippel Lindau disease gene is located on the short arm of chromosome 3 (3p25-26). Like other phacomatoses, there are no skin manifestations in VHL disease.

The he mangioblastoma of the central nervous system represents the most typical and consistent is ion of this condition [14]. The relative frequency of major lesions of Von Hippel Lindau's dis-ease is as follows:

a. Hemangioblastoma of the central nervous system: 60-80%

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b. Retinal Hemangioblastoma: 50-60%

c. Pancreatic disease: 30-65%

d. Cancers or kidney cysts: 30-60%

e. Phenochromocytome: 11-19%

VHL'S Diagnostic Criteria

The clinical diagnosis [15] is based on:

a. in the absence of family history, the presence of two hemangioblastomas or heman-gioblastoma and a visceral lesion (pheochromocytoma, multiple renal cysts, kidney cancer, cysts or pancreatic tumors, cystadenoma of the epididym);

b. with a family history, the presence of a hemangioblastoma or a visceral lesion.

Conclusion

Phacomatoses, or neurocutaneous syndrome, include a large group of genetically-cause conditions responsible for the formation of hamartomata and/or tumor lesions mainly affecting structures of ectodermal origin and mainly resulting in skin and central nervous system damage. Neurological manifestations are often a major component of the prognosis. The most common diseases are type 1 and 2 neurofibromatosis, rose tub Bourneville tuberous pink sclerus, von Hippel-Lindau disease and Sturge-Weber syndrome. Neu-roimaging, including MRI, has an essential role in positive diagnosis, prognostic evaluation, re-ferral of therapeutic management, evolutionary follow-up of lesions and family screening.

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