Case Report on: Tuberous Sclerosis

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Authors’ contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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ABSTRACT

Introduction: Tuberous sclerosis is a rare hereditary disease that creates noncancerous tumours in the brain, kidneys, heart, liver, eyes, lungs, and skin. Seizures, intellectual incapacity, developmental delays, and behavioral issues are just a few of the warning symptoms. Like Skin problems, and lung and kidney problems. A hereditary mutation in one of two genes causes TSC. TSC1 and TSC2 are two distinct TSC types. The proteins hemartin and tuberin, which act as tumour suppressors and govern cell proliferation and differentiation, are produced by these genes.

[1]

Background: Tuberous sclerosis complex (TSC) is a rare genetic disorder that affects 1 in every 6,000 to 1 in every 18,000 people. It's a life-threatening condition caused by the formation of benign tumors/lesions in several organs. Tumors can affect organ growth and/or function and are frequent in the brain, heart, skin, kidneys, and lungs. The number of organs affected, as well as the size of tumours inside each organ, varies greatly. As a result, the disease's symptoms are extremely variable and unexpected [2].

Case Presentation: A 35-year old female admitted to AVBRH on date 25/11/2021 with the chief complaint of fever chills, nausea, vomiting and itching and back skin over mouth and eye surrounding area since in 10 days.

History of Present Illness: Tuberous sclerosis, also known as tuberous sclerosis complex, is a

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rare genetic disease that causes non-cancerous (benign) tumours in the brain, skin, kidneys, heart, eyes, and lungs. A 35-year-old female admitted to hospital on date 25/11/2020 with the chief complaint was fever chills, nausea, vomiting and itching and back skin over mouth and eye surrounding area since in 4 month.

**Interventions:** The patient was treated the patient was started on Injection- Ceftriaxone, 1gm – Intravenous – BD- Antibiotic, Injection- Pan, 40mg – Intravenous- BD – Antacid, Injection- Livipril, 100mg – Intravenous- BD- Anticonvulsant, Injection- Paracetamol, 150mg – Intramuscular- SOS- Antipyretic.

**Conclusion:** During hospital stay with the chief complaint of patient are fever chills, nausea, vomiting and itching and back skin over mouth and eye surrounding area since in 10 days. Her situation was critical, therefore she was brought to AVBR Hospital and treatment was started right away.

**Keywords:** Tuberous sclerosis; fever chills; nausea; vomiting and itching and back skin over mouth and eye surrounding area.

### 1. INTRODUCTION

Tuberous sclerosis complex (TSC) is a rare autosomal dominant neurocutaneous illness that causes benign tumours to form in the brain, skin, retina, and viscera. It's marked by cutaneous abnormalities, neurologic symptoms, and the formation of hamartomas in several organs, all of which contribute to higher morbidity and mortality [3]. The most common oral symptoms are fibromas, gingival hyperplasia, and enamel hypoplasia. The treatment of these patients is frequently interdisciplinary, involving professionals from various professions. The treatment of these patients is frequently interdisciplinary, involving professionals from various professions. In the present study author discuss the clinical, radiological, and histological findings of a 35-year-old female patient with tuberous sclerosis [4].

### 2. CASE HISTORY

#### 2.1 Patient Information

The female patient 35-yearold female admitted to AVBRH on date 25/11/2021 with the chief complaint of fever chills, nausea, vomiting and itching and back skin over mouth and eye surrounding area since in 10 days. All investigations were completed after admission to the intensive care unit, including CBC, KFT, LFT, MRI, CT Scan, EEG, ECG, Vision test, skin test, developmental or psychiatric evaluation, screening, and genetic testing [5].

#### 2.2 Past Medical History

The patient was having a history- she is known case of seizures disorder since in 12 year and tuberous sclerosis disease since 2 year. And she is no history of HIV and asthma and hypertension and DM [6].

#### 2.3 Psychosocial History

Patient maintains interpersonal relationship with family, friends and relatives.

#### 2.4 Family History

A 35-year old female is a known case of Tuberous sclerosis complex. There is no other family history of illness.

#### 2.5 Socioeconomic Status

Mrs. Sunanda kavre, is the only house wife but your husband work in bread winner of the family he is electrician and works in MSEB he earns 25,000/- month approximately. They live in their own house made up of bricks and cement. There is proper electricity and water supply in their house.

#### 2.6 Nutritional Status

They consume mixed diet.

#### 2.7 Personal History

Patient is having disturbed sleeping pattern. Hygiene is well maintained.

#### 2.8 Physical Examination

#### 2.8.1 General parameter

Temperature- 101\(^0\) F, Pulse- 82 breath/ min, Respiration-22 breath/ min,
Heart sound- 100 beats/min, Blood pressure- 130/80 mmhg, Height- 167cm, Weight – 66kg, BMI – body mass index = 23.7 /M².

2.8.2 General physical examination

There were many well-defined reddish-brown sessile nodular growths in a classic "butterfly pattern" on the forehead, nose, and cheeks, as well as angiofibromas in the front and back trunk regions. Periungual fibromas or Köenen tumours were identified. In the upper and lower extremities as sessile and solid nodular growths of various diameters. A well-defined roughened hypermelanotic patch with an orange peel appearance indicative of sharpen patch was noticed in the left shoulder girdle and right lumbosacral region. The vital signs were found to be within acceptable ranges [7].

2.8.3 Intraoral examination

Similar well-defined, sessile, robust, and nodular growths of varied sizes were detected in the marginal and connected gingiva in the upper and lower anterior regions. In the lower anterior, there was also marginal and papillary gingival enlargement. On the occlusal surface of posterior teeth, several hypoplastic enamel pits were discovered. Based on the patient's history and clinical findings, a preliminary diagnosis of drug-induced gingival expansion was determined. The distinction between gingival fibromas and neurofibromas was carefully considered [8].

2.9 Radiological, Hematological, and Histopathological Investigations

There was no suggestion of bony involvement on a research aims. In the periapical area of 21, a radiopaque mass resembling a tooth with enamel, dentin, and pulp was seen, suggesting compound odontome. The left border of the ventricle was found to be displaced inferiorly on a chest radiograph, indicating left ventricular hypertrophy. Axial computed tomographic portions of the brain in the subependymal regions of both ventricles revealed hypodense patches measuring roughly 5 mm 5 mm, indicating multiple calcified tuberous lesions (subependymal nodules). There were no abnormalities seen on an ultrasound of the whole abdomen. The sedimentation rate was discovered to be increased to 32 mm [9].

2.9.1 Hematological investigations were

Except for a stretched out, hypoplastic stratified squamous epithelium overlying connective tissue stroma in an erythrocyte hematoxylin and eosin-stained portion of the intraoral tissue specimen, which revealed a stretched out, hypoplastic stratified squamous epithelium overlying connective tissue stroma in an erythrocyte hematoxylin and eosin-stained portion of the underlying connective stroma. Dense, thick collagenous bundles with hypocellularity, few fibroblasts, mesenchymal cells, restricted blood arteries, and a dense, thick connective stroma with hypocellularity made up the underlying connective tissues. The connective tissue stroma was overlain by orthokeratinized stratified epithelium in the extraoral biopsy specimen. The epithelium appeared flattened and thinned out, with an abundance of intensely stained melanocytes in the basal cells. The underlying connective tissue was made up of collagen bundles, mesenchymal cells, and a few nerve fibre bundles. There were no sebaceous glands visible [10].

2.10 Clinical Finding

- Skin Abnormalities – Present
- Seizures- Present
- Cognitive Disabilities - Absent
- Kidney Problem – Present
- Heart Issues – Absent
- Lung Problem – Absent
- Eye Abnormalities – Present

2.11 Diagnosis Test

Diagnostic tests to detect growths or tumours in the body will most likely include:

1. Magnetic resonance imaging (MRI) is a type of imaging that (MRI). This test creates detailed images of the brain or other regions of the body using a magnetic field and radio waves. Done

2. A CT scan is a type of computerised tomography (CT) scan. 2. A CT scan is a type of computerised tomography (CT) scan. The brain or other regions of the body are imaged in cross-section and sometimes 3-D using this X-ray technology. Done

3. This examination, also known as sonography, creates images of particular bodily components, such as the kidneys,
using high-frequency sound waves. Not done

4. **Echocardiogram.** Sound waves are used to create images of the heart in this exam. **Done**

5. Electrocardiogram (ECG) (ECG or EKG). This test records the heart's electrical activity. **Done**

6. Vision test - **Done**

7. skin test - **Done**

8. psychiatric or developmental evaluation- **Done**

9. Genetic testing and screening - **Done**

### 2.12 Therapeutic Intervention

General measures: To check the vital sign (Temperature pulse respiration and BP, ) airway, fluid and electrolyte balance and prevention of complications like seizures, pulmonary aspiration, pressure source, thrombo phlebitis are mandatory. Health management includes physiotherapy, health diet.

### 3. MANAGEMENT

Patients with Tuberous Sclerosis Complex are treated in intensive care units or burn centers for the most part. Since there is no particular treatment for Tuberous sclerosis complex, most patients are treated symptomatically [11,12].

#### 3.1 Medical Management

- **Injection-** Ceftriaxone, 1gm - Intravenous – BD-Antibiotic
- **Injection-** Pan, 40mg – Intravenous- BD – Antacid
- **Injection-** Livipril, 100mg – Intravenous- BD-Anticonvulsant
- **Injection-** Paracetamol, 150mg – Intra muscular-SOS- Antipyretic

#### 3.2 Treatment

- **Medication.** To control seizures, anti-seizure drugs can be prescribed. Some drugs can be used to treat cardiac arrhythmias, behavioural issues, or other symptoms. Ever olimus (Afinitor, Zortress) is a drug that can be used to treat many forms of brain and kidney tumours that can't be removed surgically. A medication called sirolimus, which comes in the form of a topical ointment, can be used to treat acne-like growths of masses.

- **Surgical procedure.** If a growth is interfering with an organ's ability to function, such as the kidney or heart, it can be surgically removed. Seizures triggered by brain growing that don't respond to medication may often be controlled with surgery. Skin growths can be improved by surgical procedures such as dermabrasion or laser therapy.

- **Various types of therapy.** Occupational, physical, and speech therapy, for example, can assist children with tuberous sclerosis increase their capacity to perform everyday jobs and activities.

- **Educational and vocational services.** Children with developmental disabilities and behaviour problems will benefit from initial intervention and special needs programmes to help them adjust to school and reach their full potential. Social, vocational, and recovery programmes can be continued throughout one's life if necessary.

- **Psychological and behavior management.** Talking to a mental health professional will help children understand and adapt to their condition. A mental health professional may also assist with behavioral, psychological, or emotional problems, as well as suggest support [13].

### 3.3 Tuberous Sclerosis Treatment

There is no specific treatment for the Tuberous sclerosis

1. With the help of the medicine we can control the growth of the tumour.
2. With laser treatment we can manage a small growth of the tumour
3. Brain tumours can be treated with medicine.
4. We can manage the tumours in the kidney with the help of the surgical management.

### 3.4 Nursing Management

#### 3.4.1 Nursing assessment

Tuberous sclerosis is a chronic disease that necessitates close supervision and follow-up due to the fact that certain warning signs will take time to manifest. Tests similar to those performed during diagnosis can be included in a routine of daily follow-up monitoring during life.
Complications can be avoided if issues are identified early.

### 3.4.2 Coping and support

1. When a patient is diagnosed with tuberous sclerosis, the patient's family may be confronted with a number of challenges and unknowns. The inability to forecast how a patient's health and growth will progress over time is one of the most difficult features of this condition.

2. The patient can only have minor issues and perform similarly to peers in terms of academic, social, and physical abilities. Alternatively, the patient's health and developmental concerns may be more severe, resulting in a life that is less autonomous or different from what the patient anticipated [14].

### 3.4.3 Teaching about the coping strategies to the patient and relatives

1. Make a timetable for screenings. Learn what you can about tuberous sclerosis, and work closely with your doctor to build a routine for screening and tracking health and developmental issues. Infections can be avoided by detecting and handling problems early on.

2. Initialization of the treatment with behavioural issues as soon as possible. Tuberous sclerosis can cause behavioural problems in children, which can be difficult for parents to deal with. And it is not the fault of the patient. When problems emerge, consult the patient's psychiatrist and work with the school or a mental health expert to address the curriculum as well as behavioural and mental health services. The earlier patients are seen, the better.

3. Show love and support to others. Love and support for patients are critical in assisting them in reaching their full potential. Counselling with a mental health provider, if necessary, will assist with adjustment and coping. Parents may find respite care services to be beneficial.

4. Make friends with other families. Connecting with other families dealing with tuberous sclerosis can be beneficial to the patient. Inquire about a local support group with the patient's health care provider, or contact the Tuberous Sclerosis Alliance for further information [15].

### 3.4.4 Preparing for appointment of the patient

1. Tuberous sclerosis symptoms and signs may be detected as early as birth. Alternatively, the patient can first discuss his or her issues with the doctor. Following a physical examination, the patient may be referred to one or more physicians for further testing and care.

2. Request that the patient be accompanied to the appointment by a trustworthy family member or friend. Bring someone along who can offer moral support and assist the patient in remembering everything [16].

### 4. NURSING DIAGNOSIS

1. Hyperthermia related to infection, and secondary to disease condition.

2. Excessive fluid volume related to fluid accumulation in the body and secondary to disease condition.

3. Imbalanced nutritional pattern less than body requirement related to nausea and vomiting.

4. Skin integrity and skin rashes related to disease condition.

5. Acute pain related to tuberous sclerosis in face and eyes [17].

### 4.1 Collaborative Problems/ Potential Complications

- Collection of the fluid in the brain interstitial spaces
- Cardiac complication
- Renal damage
- Dysfunction of the Lung
- Increase risk of (malignant) tumors.
- Visual damage

### 5. EVALUATION

#### 5.1 Expected Patient Outcomes

Expected patient outcomes may include:

#### 5.1.1 Demonstrates tolerance for increased activity

1. Describes adaptive methods for normal work.
2. Stops all activities which cause intolerance symptoms
3. Keeps vital signs within target range (pulse, blood, breathing rate and pulse oximetry).
4. Identifies and measures to prevent factors that contribute to activities intolerance
5. Set priorities for activities
6. Plans activities for energy conservation and fatigue and dyspnea reduction

5.1.2 Maintains fluid balance
1. Peripheral and sacred edema decreased in exhibits
2. Demonstrates methods in preventing edema

5.1.3 Is less worried
a. stop unpleasant circumstances
b. Sleeps at night comfortably
c. Reports decreased stress and anxiety

5.1.4 Makes decisions regarding care and treatment
a. States ability to influence outcomes

5.1.5 Adheres to self-care regimen
a. Performs and records daily weights
   Ensures dietary intake includes no more than 2 to 3 g of sodium per day [18].

6. COMMUNICATION
Determine how difficult it is to communicate or respond to a query using words. Encourage the patient's communication efforts. Speak slowly and clearly, and seek advice from speech therapists [18].

7. DISCUSSION
TSC is a difficult condition to treat. Nurses may provide education about its multisystem manifestations, adequate required monitoring, and strategies to successfully follow a care plan in collaboration with patients and families. Epilepsy treatment must be appropriate and efficient. Novel treatments, such as mTOR inhibition, have the potential to enhance seizure control, as well as many other TSC symptoms and quality of life. More research is required on how to effectively manage side effects in order to improve tolerability and better understand the role of these novel therapies in TSC patients [1].

8. CONCLUSION
The patient was admitted to hospital with the chief complaint of fever chills, nausea, vomiting and itching and back skin over mouth and eye surrounding area since in 10 days and his condition was very critical and the patient was admitted in AVBR Hospital, immediate treatment was started by health team member and all possible treatment were given and now the patient condition is satisfactory [19].

9. STRENGTH
Patient was 35 year female patient tolerate all the medication and well response within seven days to the therapeutic treatment of the hospital which was given as a treatment.

INFORMED CONSENT
Before taking this case, information was given to the patients and their relatives and Informed consent was obtained from patient as well as relatives.

ETHICAL APPROVAL
we conducted our research after obtaining proper IEC approval.

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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