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Southeast Asian Journal of Case Report and Review

Journal homepage: https://www.sajcrr.com/

Case Report

Case study on 19-year-old female with Mc-Cune Albright syndrome

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ARTICLE INFO

Article history:
Received 15-10-2023
Accepted 02-11-2023
Available online 20-11-2023

Keywords:
McCuneAlbright syndrome
Genetic mutations
Fibro adenoma
Mastectomy
NSAIDS

ABSTRACT

McCune-Albright syndrome is a genetic condition that affects your skin, bones and endocrine system (endocrine organs). It causes scars on bones, skin pigmentation and elevated function of growth-regulating hormonal glands. This can be mild in few people and quite severe to life-threatening in others. This syndrome can affect any age group of people like children, adults and old population due to some genetic mutations. These mutations can be sudden and unpredictable. We have come across a rare case called McCune-Albright syndrome where a 19-year-old female presented with chief complaints of lump in left breast associated with pus discharge sine a week. There have been changes in the skin and there is no history of retraction of nipple and any swelling in axilla, neck and opposite breast. Patient has gone to the doctor when she found a lump one month back, doctor has advised some of the diagnostic tests but she completely ignored and after one month she came to the hospital and has been admitted in the general ward. She was treated with vitamin supplements, antibiotics, antacids, NSAIDS.). The surgery procedure is simple mastectomy of left breast. This is done due to the fibro adenoma present in left breast. The patient was recovered in 10 days.

1. Introduction

McCune-Albright syndrome is a genetic condition that affects your skin, bones and endocrine system (endocrine organs). It causes scars on bones, skin pigmentation and elevated function of growth-regulating hormonal glands. This can be mild in few people and quite severe to life-threatening in others. This syndrome can affect any age group of people like children, adults and old population due to some genetic mutations. These mutations can be sudden and unpredictable.[1,2]

2. Etiology/Pathophysiology

The main cause of McCune Albright is the mutation od a gene called GNAS. GNAS is gene that is used for the protein synthesis responsible for hormonal activities. It provides instructions for making a part of a protein complex called a guanine nucleotide-binding protein or G protein. During signal transduction process, the G proteins activates a complicated network of signal pathways that impacts many cell functions by regulating hormonal activity. The protein obtained from the GNAS gene can stimulate the activity of an important enzyme called “adenylate cyclase”. When this gene mutates, adenylate cyclase can be constantly in an activated state. Because of this continuous activated state, it leads to over hormonal release from several hormonal glands which can ultimately results in the unwanted growths on the bones, skin and other tissues

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https://doi.org/10.18231/j.sajcrr.2023.021

and causes symptoms of McCune Albright. As the mutated gene leads to constitutive activation of the alpha subunit, it causes elevated intracellular c AMP and starts mediating the action of downstream hormones. This increased c AMP signalling leads to clinical manifestations of the syndrome. In bones, the elevated cAMP causes the osteoblasts to get differentiated into the stromal cells and also inhibits further differentiation of the cells, resulting in fibrous dysplasia (abnormal extra growths on the bone). In skin, it results in the over stimulation of melanin synthesis (discoloration of the skin). In endocrine tissue, it leads to elevated production and secretion of a specific hormones. The effected endocrinal organs include gonads, thyroid gland, pituitary gland, parathyroid gland, and adrenal glands. The characteristics of the fibrous dysplasia is an elevated number of thin and irregular trabeculae. The bone marrow spaces are also get replaced by the fibroblastic tissue. These lesions may be monostotic (involving a single bone) or polyostotic (involving multiple bones). They are commonly identified through radiograph imaging studies, fibrodysplasia shows radiolucent, ground-glass pattern of tissue due to the defective mineralization of the bones.

2.1. Diagnosis

Full endocrinal studies are performed like measuring Gonadotropins (luteinizing hormone [LH], follicle-stimulating hormone [FSH]) and sex hormone levels) and to evaluate various endocrinal organs functioning and their current status. Genetic testing is also done to identify any abnormal gene mutations like biopsy of skin, bone, muscle, thyroid and other tissues. Radiological imaging studies like CT scan, X ray and MRI scanning of the body used to rule out extra growths on the bones or organs. Thyroid levels should be checked (thyroid-stimulating hormone [TSH], thyroxine [T4], antithyroid antibodies). Growth hormone (GH) and insulin like growth factor 1 (IGF-1) levels also to be checked. ABG determination can be helpful in suspecting any signs of acidosis. Gastrointestinal endoscopy is used identify and polyps in the GI tract.[3–6]

3. Treatment

The treatment is unique for each patient diagnosed with the McCune-Albright syndrome. There is no ultimate cure for this syndrome and treatment mainly focuses on the symptoms of the patient. Aromatase inhibitors can reduce the levels of estrogen by inhibiting the activity of an enzyme called aromatase. Because, estrogen can act as a factor for inducing growth of cancerous cells in the breast. Bisphosphonates are used to reduce the risk of bone fractures and to treat extra bone growths. They also help in relieving pain in majority of the cases. To treat hyperthyroidism, pharmacologic agents like thionamides, methimazole, radioiodine. To treat hypogonadism - propriate hormone replacement therapy is given. For hypophosphatemia with hyperphosphaturia, aggressive oral phosphorus replacement is given. Surgical interventions are considered like oophorectomy for precocious puberty. Traction and fixation for bone fractures or any lesions. For uncontrollable hyperthyroidism, thyroidectomy (partial or total). Surgical removal of the offending lesions is done for gigantism or acromegaly conditions.

4. Case Presentation

4.1. History of present illness

A 19-year-old female patient present in general ward with chief complaints of lump in left breast associated with pus discharge since a week. Patient was. Asymptomatic apparently one month back then she has developed lump in left breast which is insidious in onset and is gradually progressive in size. Initially the size of lump was in a grape shape now increased to progressive size. There has been changes in the skin (discolouration of the skin from shoulder to left breast since 15 days and it is increasing progressively) and there is no history of retraction of nipple, swelling in axilla, neck and opposite breast. Patient has gone to the doctor when she found a lump one month back, doctor has advised some of the diagnostic tests but she completely ignored because of her poor economical status and after one month she came to the hospital and has been admitted in the general ward.[6–8] She has complaint of lower back pain and weight loss in last 15 days, there is no history of chest pain, shortness of breath or trauma. She has no complaint of pain in the breast region.

4.2. Social history

She is single unmarried women and she is doing her under graduation.

4.3. Allergies

No known food, medicine and environmental allergies.

4.4. Past medical history

She has no significant history of similar complaints in past and no known history of hypertension, diabetes, tuberculosis, asthma and thyroid.

4.5. Surgical history

She had lower limb fracture and open reduction internal fixation (ORIF) was done at 14 years of age.

4.6. Menstrual history

Attained menarche was at 14 years of age, last menstrual period was regular and menstrual cycles were regular and menstrual flow was normal.

4.7. Medications history

Currently no medications are being used.

4.8. Birth history

1. Patient was third child and she has under weight-2.5kg, cry-positive.
2. Normal vaginal delivery and no NICU admission.
3. Immunization as per schedule.

4.9. Family history

1. Her elder brother was developmentally delayed.
2. Her mother marriage was consanguineous.

4.10. Physical examination

Vitals: Temperature-98.6F, Blood pressure-100/60mmHg, Heart rate-72/min, Respiratory rate-22/min, body mass index-21.8, GCS- E4V5M6, GRBS- 96mg/dl, SPO2-96@room air.

4.11. General examination

She has appeared pale and anxious; she is cooperative, and moderately built and nourished.

4.12. Local examination

4.12.1. Inspection

1. Right breast appears normal, left breast appears asymmetrical and different position.
2. Nipple- no retractions, no fissures present, no discharge
3. Upper quadrant and lower quadrant have approximately a 4x3cm lump, surface smooth, margin regular.
4. Skin over the left breast pigmented with pus drainage opacities

4.12.2. Palpation

1. Right breast normal, left breast tenderness positive.
2. Lump present in upper and lower quadrants.

4.12.3. Respiratory function

She has normal respiratory rate that is 22/min.

4.12.4. Cardiovascular

She has heart rate which is regular with rhythm and there were no murmurs, wheezing sounds.

4.13. Gastrointestinal

She has per abdominal region soft and no extra growths and abnormalities were found.

Bowel and bladder was normal and appetite was normal.

5. Initial Evaluation

5.1. Laboratory studies

Initially laboratory investigations were done to the patient in the general ward and revealed that she has less amount of haemoglobin level of 8gm% and that revealed she has anaemia and she has normal count of platelets that doesn't include the case of thrombocytopenia. For complete blood picture she had impression of Anisopoikilocytosis which refers to a condition of varying sizes and shapes of erythrocytes, hypochromic anaemia, microcytosis macrocytes. She has elevated alkaline phosphatase levels that is 941U/L. Other values were normal in liver function test and renal function test was normal.[9]

1. *Chest x-ray:* Bilateral lung fields were normal, bony ribcage were normal, soft tissue shadow was normal.
2. *Viral markers:* Negative.
3. *ECG:* Tachycardia.

6. Histopathology/Cytology Report

6.1. Nature of the specimen

Specimen of trucut biopsy of left breast swelling.

6.2. Gross appearance

Received scanty grey white material, linear soft tissue bits.

6.3. Microscopic apperance

Sections from tiny bits revealed numerous ducts lined by cuboidal epithelium with areas of stratification, epitheliosis and foci of well formed papillary structures with fibrocollagenous core, few with cystic dilatation, cyst macrophages and foci of apocrine metaplasia, separated by scant stroma.

6.4. Impression

F/S/O: Ductal hyperplasia, advised wide excision.

6.5. USG of left breast

Upper outer and lower outer quadrant have approximately a 4x3cm lump, surface smooth, margin regular, skin over breast pigment with pus draining opacities, no alterations in the position seen on raising. Right breast is normal, local raise of breast, tenderness positive.

6.6. Differential diagnosis

1. Neurofibromatosis
2. Osteofibrous dysplasia
3. Non-ossifying fibromas
4. Idiopathic central precocious puberty
5. Ovarian neoplasm

6. Fibrous dysplasia
7. Cafe-au-late-spots
8. Extra medullary acute myeloid leukaemia
9. Mastitis
10. Costochondritis
11. Intracystic papilloma
12. Atypical ductal hyperplasia
13. Ductal carcinoma in situ
14. Papillary carcinoma
15. Fibroadenomas

7. Confirmatory Evaluation

Histopathological studies were done, the specimen of trucut biopsy of left breast swelling was the nature of the specimen and the appearance was scanty grey white material and has linear soft tissue bits and the microscopic appearance was the sections from tiny bits revealed numerous ducts lined by cuboidal epithelium with areas of stratification, epitheliosis, foci of well-formed papillary structures with fibro collagenous core and few with cystic dilatation, cyst. macrophages, foci of apocrine metaplasia separated by scant stroma. And it confirms the sign of *Ductal Hyperplasia* and wide excision was advised.

7.1. Diagnosis

Based on the signs and symptoms and laboratory findings a diagnosis of *Mc-Cune Albright Syndrome* was made.

8. Management

Mc-Cune Albright syndrome symptoms occur together as a group or it can be a set of conditions characterised together, the treatment cannot be the ultimate cure because it is syndrome. The symptomatic treatment is to be given for the relief for some days. Initially when the patient has been admitted in the hospital they started vitamin supplements, antibiotics (Augmentin 625mg/tid), antacids, NSAIDS (diclofenac 50mg/bid). One pint of PRBC transfusion was done, then surgery was planned pre operative instructions are given to the patient nil by mouth, antibiotics(cephalosporins), inj.TT was given, general anaesthesia was given, anti-anxiety(benzodiazepines) were given, laxatives (Dulcolax) was given. Part preparations were done. Secured green (18G) cannula on right hand. Shifted to modular operating theatre (MOT). The surgery procedure is simple mastectomy of left breast. Indication was fibro adenoma present in left breast, anaesthesia type-general anaesthesia, and incision-elliptical incision nipple areola complex. intraoperation findings were heterogenous tissue with no evidence of normal beast tissue noted and the procedure here is under strict aseptic precaution. Patient is in supine position under general anaesthesia, parts painted and draped, elliptical incision was made and deepened till the lump in the left breast was visualised then the lump is separated from the rest of the tissue in same plane from all the directions lump is separated and removed intact, haemostasis secured, sub-cutaneous suturing was done using catgut skin sutured with prolene. Procedure uneventful, patient stable throughout the procedure. After the surgery was done nil by mouth was advised till the further orders, IV fluids were recommended that is 2-pint ringer lactate, 2-pint 5% dextrose, 2-pint dextrose normal saline, antibiotics (cephalosporins, aminoglycosides), antacids, painkillers and vitals should be monitored accordingly. On post operation day-1 the vitals were normal, again one pint of PRBC transfusion was done, sips of liquid were allowed and vitamin supplements were added. On post operation day-2 the vitals were normal, diet was advised and patient has complained cough, syrup ambroxol was prescribed. Same drugs were indicated till the discharge of the patient. Again, 1pint of PRBC transfusion was done on post operation day-6 because of low haemoglobin levels (8gm %).

8.1. Prognosis

Prognosis could be challenging to ascertain due to the varying severeness of the disease. Generally, younger population are at a greater risk for this syndrome. The major treatment involves surgical procedures like mastectomy, fixation of bones. Malignancy of the abnormal growths is a rare type of presentation.

9. Enhancing Healthcare Team Outcomes

The main point in this is that how the healthcare team works in order to bring a proper outcome with the diagnosis. Here the staff works accordingly coordinate with each other to achieve proper diagnosis in order to give proper treatment for concerned diagnosis. The staff who is mainly involved are doctors, duty medical officers, nursing staff, technicians these all play major role in the probability of making correct diagnosis for the patient. There should be communication between them if there is no communication it leads to improper diagnosis. Any alterations in the middle of making diagnosis the discussion should be made and changes are to be done. Communication here plays a vital role.

10. Discussion

McCune-Albright syndrome is a genetic condition that affects your skin, bones and endocrine system (endocrine organs). It causes scars on bones, skin pigmentation and elevated function of growth-regulating hormonal glands. This can be mild in few people and quite severe to life-threatening in others. This syndrome can affect any age group of people like children, adults and old population due to some genetic mutations. These mutations can be sudden and unpredictable.

11. Conclusion

Mc-cune Albright syndrome is a genetic condition that effects the skin, bone and endocrine system. In our case report the patient effected with the skin and endocrine system. The causes are still unknown. These cases are treated symptomatically as it is a syndrome.

12. Source of Funding

None.

13. Conflict of Interest

None.

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Cite this article: Mannem A, Janampeta A, Kota K, Rachakonda S, Undru S, Chillara T. Case study on 19-year-old female with Mc-Cune Albright syndrome. *Southeast Asian J Case Rep Rev* 2023;10(4):94-98.