Case Report

ROTHMUND THOMSON SYNDROME

1THAMER MOHAMMAD BSOUŁ
2TALAL NSER AL-RAWASHDEH
3MUE’N MAHMOOD AL-WESHAAH
4ENAS FAWAZ OTHMAH
5AYMEN DAOUD SHAWEKH

ABSTRACT

Rothmund Thomson Syndrome is a rare autosomal recessive syndrome caused by homozygous or compound heterozygous mutations in (RECQL4) gene.

It was first described in 1868 by a German ophthalmologist (Rothmund1) and later in 1936 by an English dermatologist (Thomson2) who reported another three similar patients but the oponym Rothmund Thomson Syndrome (RTS) was named by Taylor3 in 1957.

SIGNS AND SYMPTOMS

The first most frequent sign is poikiloderma in addition to sparse scalp hair and eyelashes, short status, skeletal abnormalities, juvenile cataract and premature aging.

There are two subtypes of the syndrome; RTS Type 1 and RTS Type 2 where only type 2 has high risk of osteosarcomas in childhood and skin cancer later.

DIAGNOSIS OF RTS

Diagnosis is difficult and usually not definite but according to (Wang and Plon) it can be diagnosed if poikiloderma or atypical rash were found in infancy in addition to at least two of the following signs:

− Sparse hair on scalp and eyebrows
− Short status
− Congenital bone defects
− Dental abnormalities such as unusual crown formation, microdontia, hypodontia, short roots (rhizomicry) and connective tissue disease of the gingiva.
− Dystrophic nails.
− Cataract.
− Esophagus and pyloric stenosis that causes feeding problems in infancy which may lead to aplastic anemia or leukopenia.

PROGNOSIS AND TREATMENT

Although some signs of early aging are usually seen but life span of the patients is not altered provided that neoplastic complications do not occur and the patients should be managed by a team which includes dermatologist, ophthalmologist, orthopedic surgeon, oncologist and dentist.

CASE REPORT

A 42-year old female attended dental clinic complaining of grade three mobility of all her remaining teeth and roots which were hypoplastic in general and previously extracted 19 of her teeth for the same reason with moderate to good oral hygiene.

Upon examination the patient was of short status of 120 cm, her weight was 39 kg. she had marked telangiectasia on her face and both limbs especially at the extensor surfaces with multiple cafe-au-lait spots on her neck, shoulders and upper limb. She appeared to have little scalp and eyebrows and lashes hair, her nails were obviously dystrophic hypotrophic with multiple ridging formations.

After taking detailed history the patient informed us her elder sister and her son have nearly the same symptoms, and by reviewing her medical file we found that she was diagnosed to have cataract in both eyes with barred vision and she had undergone two surgeries to enlarge the esophagus as she was diagnosed
Rothmund Thomson Syndrome

Fig 1: Dystrophic nails

Fig 2: Poikiloderma

Fig 3: Juvenile cataract and sparse eye lashes

Fig 4: Sparse hair of the scalp

Fig 5: White lesion (Leukoplakia)

Fig 6: Ulcerative leukoplakia

Fig 7: Dentures made

Fig 8: Dentures provided
to have pyloric stenosis; the first surgery was done at the age of eleven while the second one was at the age of thirty. She made regular visits to dermatologist as she complained of severe skin dryness in addition to her need to relief the pressure made by fibrous tissue around her fingers.

Previous blood tests revealed microcytic hypochromic anemia with 5.6 haemoglobin ratio before pyloric stenosis surgery.

**DIAGNOSIS**

A definite diagnosis of RTS was not available due to non specific symptoms, but after reviewing time onset of the symptoms especially the skin manifestations and its' association with other symptoms of sparse scalp and eyebrows' hair, short status, cataract, nail and teeth abnormalities; she is most likely to have RTS. RECQL3, 4, 5 test was recommended since two thirds of the patient have RECQL4 mutation but the patient didn’t agree to do the test.

Blood tests were made after she was diagnosed and they revealed improved haemoglobin ratio 12.4 with normal full chemistry except high cholesterol level.

Brain M.R.I was done as she complained of chronic severe headache with no obvious reason that started 3 months before but M.R.I result was normal with no malignant changes except for parasinusitis and was referred to the ENT specialist for treatment.

**TREATMENT PLAN**

The patient was informed that her remaining teeth and roots have to be removed in order to make an immediate complete denture.

Extractions were done and impressions for immediate denture were made.

While taking the impressions, we had to modify the smallest stock tray size to fit her very small mouth opening and ridges since her lips and face skin was very tight.

Immediate dentures were made and used for the next three months with soft lining material which was changed several times.

Three months later, the patient came to change her immediate dentures for definite ones but upon examination, her palate and right cheek to the level of the retro molar pad area of the mandible and the corner of the mouth were covered with thick hypokeratosed white lesion that had developed within three months.

Biopsy of the white lesion (leukopenia) was made to exclude any malignant changes which are very common in RTS type 2 patients.

The biopsy result was leukoplakia with no malignant change and she was given cortisone for one month.

After taking the cortisone for one month, the lesion improved but didn’t cure, so she was informed to do another biopsy six months later.

The definite denture was fabricated with permanent soft acrylic material (velloplast). The patient is satisfied and is doing well.

**REFERENCES**