Dyskeratosis congenita (DC), also known as Zinsser-Engman-Cole syndrome, is an inherited skin condition with the classic triad of dysplastic nails, lacy reticular skin pigmentation, and oral leukoplakia as skin changes. Individuals with DC have a high risk of progressive bone marrow failure, pulmonary fibrosis, liver disease, cancer, stenosis of the esophagus, urethra, lacrimal ducts, and avascular necrosis of the hips or shoulders. This syndrome is closely related to defects in telomere biology which is also considered a possible precedent for various psychiatric disorders. A recent pilot study has reported higher cooccurrence of neuropsychiatric disorders in pediatric and adult population afflicted by this syndrome. Although there are a couple of reports of DC with comorbid schizophrenia, no reports describing mood disorder in DC exist. With this background, we are reporting a case of an adult male with DC and comorbid psychotic and mood disorder.

**Abstract**

Dyskeratosis congenita (DC) is a rare heritable skin disorder with progressive bone marrow failure. Psychiatric manifestations could also be a presentation of this rare skin condition. Although it has been associated with psychiatric manifestations, there is a dearth of information regarding mood symptoms in this condition. We report a 41-year-old male who had presented with predominant psychotic followed by affective symptoms and was diagnosed with DC and comorbid organic delusional and mood disorder. He was also worked up for other physical manifestations of DC.

**Keywords:** Dyskeratosis congenita, organic delusional disorder, organic mood disorder, psychodermatology, telomere shortening

**INTRODUCTION**

Dyskeratosis Congenita (DC), also known as Zinsser-Engman-Cole syndrome, is an inherited skin condition with the classic triad of dysplastic nails, lacy reticular skin pigmentation, and oral leukoplakia as skin changes. Individuals with DC have a high risk of progressive bone marrow failure, pulmonary fibrosis, liver disease, cancer, stenosis of the esophagus, urethra, lacrimal ducts, and avascular necrosis of the hips or shoulders. This syndrome is closely related to defects in telomere biology which is also considered a possible precedent for various psychiatric disorders. A recent pilot study has reported higher cooccurrence of neuropsychiatric disorders in pediatric and adult population afflicted by this syndrome. Although there are a couple of reports of DC with comorbid schizophrenia, no reports describing mood disorder in DC exist. With this background, we are reporting a case of an adult male with DC and comorbid psychotic and mood disorder.

**CASE REPORT**

A 41-year-old male presented to the outpatient services with a 4-month history of psychiatric symptoms. The initial few weeks of presentation were characterized by overvalued ideas of persecution and reference, which acquired a delusional character in the subsequent months. In addition, about a month before admission, there was elevated mood, increased speech, decreased need for sleep, and abusive and assaultive behavior. Previously, he was receiving treatment and had developed QTc prolongation on electrocardiogram while on a combination of tablet haloperidol 7.5 mg, tablet chlorpromazine 400 mg, tablet aripiprazole 10 mg, and tablet valproate 750 mg. Hence, he was referred to our center for further management.

There was a history of unnatural death due to brain tumor in elder brother who had skin pigmentation anomalies. There was no significant personal history including substance use. Following admission, on general physical examination, he was having dry skin with lacy reticular pigmentation and oral leukoplakia as demonstrated in Figure 1a and b, respectively. He was also having dystrophic nails and conjunctival congestion. His body mass index was high (27.04 kg/m²). Cognitive abilities of the index case were preserved. Baseline blood biochemistry and hematological workup were unremarkable [Table 1].

**How to cite this article:** Chandrasekaran V, Sathyarayanan G, Menon V, Bharadwaj B. A case report of dyskeratosis congenita associated with both psychosis and mood disorder. Int J Adv Med Health Res 2018;5:75-7.

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| DOI: 10.4103/IJAMR.IJAMR_5_18 | |
He had a QTc interval of 473 ms.

We made a diagnosis of unspecified psychosis and mood disorder (mania) as psychotic symptoms occurred before the onset of mood symptoms and both the symptoms had an independent course but resolved together. This diagnosis was arrived based on the consensus of three qualified psychiatrists. However, the possibility of organicity was also considered in view of atypical features such as late age of onset, psychopathology, and response to treatment.

He was initiated on tablet valproate and tablet clonazepam which was gradually up-titrated to 1600 mg and 2 mg, respectively. Following cardiology liaison for QTc prolongation, tablet risperidone was initiated at 2 mg and up-titrated to 8 mg slowly over a period of 4 weeks. Alongside, lorazepam 8 mg/day was added to control manic excitement. Dermatology opinion was sought for skin pigmentation and nail changes. The index case was diagnosed with DC syndrome based on the observation of the classical triad.

Psychiatric diagnosis was revised to organic delusional and affective disorder because of the possibilities of psychiatric manifestation in DC syndrome. On evaluation for other manifestations of the syndrome, he was found to have early changes of interstitial lung disease on high-resolution computed tomography of thorax [Figure 2]. Noncontrast computed tomography of the brain revealed a normal study. The index case improved significantly over a period of 6 weeks with treatment and has been on regular follow-up since then for the past 4 months.

**DISCUSSION**

The present case had an atypical course characterized predominantly by psychotic symptoms in the initial part followed by affective symptoms with skin and nail changes suggestive of DC, a rare heritable skin condition with neuropsychiatric symptoms. In neuropsychiatric disorders, the psychiatric syndrome usually presents either as the primary manifestation or occurs concomitantly with the underlying disease as it progresses. Hence, there might be a delay in the diagnosis of the underlying neuropsychiatric condition if it primarily presents with psychiatric syndrome. The only study to date that aimed to evaluate the frequency and typology of neuropsychiatric manifestations in DC found that 50% of pediatric and 75% of adult sufferers present with primary psychiatric manifestations such as delusions and hallucinations as in schizophrenia.\(^1\) In this report, the individual primarily presented with psychiatric

| Panel                  | Investigation          | Values (normal range) |
|------------------------|------------------------|-----------------------|
| Sugar profile          | Random blood sugar     | 115 (75-140 mg/dL)    |
| Lipid profile          | Total cholesterol      | 160 (0-200 mg/dL)     |
|                        | Triglycerides          | 97 (0-150 mg/dL)      |
|                        | HDL cholesterol        | 22 (>40 mg/dL)        |
|                        | LDL cholesterol        | 119 (0-100 mg/dL)     |
|                        | VLDL cholesterol       | 19 (0-30 mg/dL)       |
| Renal function tests   | Urea                   | 18 (15-40 mg/dL)      |
| Electrolytes           | Na⁺                    | 136 (135-145mEq/L)    |
|                        | K⁺                     | 3.5 (3.5-5 mEq/L)     |
|                        | Cl⁻                    | 100 (96-106 mEq/L)    |
|                        | Calcium                | 8.5 (8.7-10.2 mg/dL)  |
|                        | Inorganic phosphorus   | 2.8 (2.5-4.3 mg/dL)   |
|                        | Magnesium              | 2.0 (1.5-2.3 mg/dL)   |
| Liver function tests   | AST                    | 45 (40-60 IU/L)       |
|                        | ALT                    | 28 0-45 IU/L          |
|                        | ALP                    | 126 (30-120 IU/L)     |
|                        | Total protein          | 8.0 (6.3-8.3 g/dL)    |
|                        | Albumin                | 4.0 (3.5-5.5 g/dL)    |
|                        | Bilirubin total        | 1.2 (0.4-1.2 mg/dL)   |
|                        | Bilirubin direct       | 0.4 (0.1-0.4 mg/dL)   |
| Hemogram               | HGB                    | 12.3 (12.2-17.3 gm/dL)|
|                        | RBC count              | 3.82 (4.3-5.7×10⁶/microL)|
|                        | HCT                    | 37.1 (38-50%)         |
|                        | MCV                    | 97.1 (80-100 FL)      |
|                        | MCH                    | 32.2 (27-32 pg)       |
|                        | MCHC                   | 33.2 (30-36 g %)      |
|                        | Reticulocyte count     | 1% (0.8-2.3%)         |
|                        | WBC count              | 8.47 (4.0-11.0×10⁶/microL)|
|                        | Neutrophil             | 61 (40-75%)           |
|                        | Eosinophils            | 4 (1-6%)              |
|                        | Lymphocytes            | 26 (20-45%)           |
|                        | Monocytes              | 9 (2-10%)             |
|                        | Platelet               | 141 (150-450×10³/microL)|
|                        | RBC                    | Normocytic normochromic|
|                        | WBC                    | Normal counts with few activated lymphocytes |
|                        | Platelets              | Adequate              |
| Thyroid function test  | TSH                    | 1.13 (0.35-5.5 µIU/mL)|
| Serology               | HIV 1/2 test           | Negative for HIV 1/2 antibodies by rapid test |
|                        | VDRL                   | Nonreactive           |
|                        | HBs Ag                 | Negative              |

\[^1^\] TSH=Thyroid Stimulating Hormone, HDL=High-density lipoprotein, LDL=Low-density lipoprotein, VDRL=Very low-density lipoprotein, ALP=Alkaline phosphatase, AST=Aspartate transaminase, ALT=Alanine transaminase, WBC=White blood cell, RBC=Red blood cell, MCV=Mean corpuscular volume, MCH=Mean corpuscular hemoglobin, MCHC=Mean corpuscular hemoglobin concentration, HGB=Hemoglobin, HCT=Hematocrit.
We acknowledge that lack of objective tests to confirm our finding as a major limitation, and it is hard to conclude a link between the disorders in a single case report. Hence, we conclude that major psychiatric disorders with atypical presentation should raise a possibility of an organic etiology as it could aid in timely intervention and minimizing morbidities.

Further long-term studies with larger sample size are required in such specific populations to understand the course and the varied clinical presentations. Furthermore, there is a need to study the association between major psychiatric disorders and telomere length in drug naïve patients and the changes with treatment.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published, and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

**Financial support and sponsorship**

Nil.

**Conflicts of interest**

There are no conflicts of interest.

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**Figure 2**: High-resolution computed tomography of thorax showing early interstitial changes.