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Clinical Image

Dyskeratosis Congenita with Pulmonary Fibrosis

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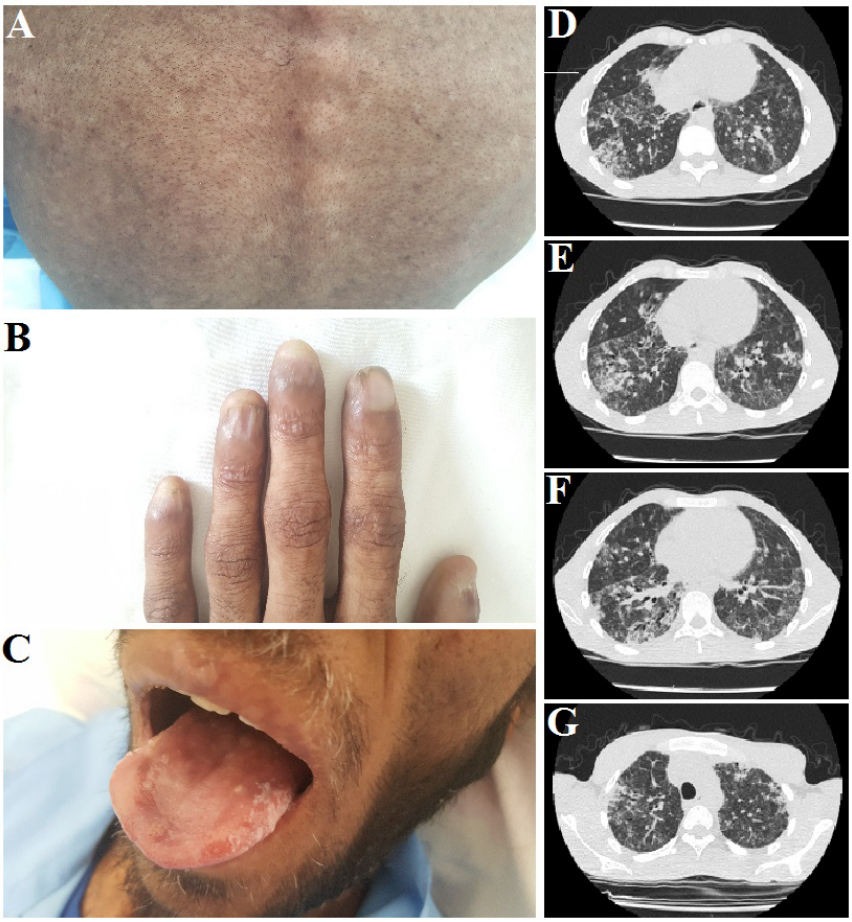

Figure 1: A) Cutaneous examination revealed hyper-pigmented and hypo-pigmented macules in a reticulated pattern all over the skin, especially in upper trunk. B) Nail dystrophy and pterygium. C) Tongue was eroded, with marginal small white irregular patches (leukoplakia). D,E,F and G) Computed tomography (CT) of his chest revealed inter lobular septal thickenings in both lungs' peripheral sites as 'honey combing' pattern which is consistent with pulmonary fibrosis.

Medical Image

A 19-year-old man presented to pulmonary clinic with a 3-month history of progressive persistent dyspnea and dry cough. Six years earlier, he had had Idiopathic Thrombocytopenic Purpura and subdural hematoma. Since then, he took 10 milligram of Prednisolone and 75 milligram of Azathioprine daily. Physical examination revealed tachycardia (110/min) and tachypnea (35/min). His Oxygen saturation was 76% in ambient air and 90% with oxygen supplementation (five lit/min, nasal cannula). Cutaneous examination revealed hyper-pigmented and hypo-pigmented macules in a reticulated pattern all over the skin, especially in upper trunk (Panel A); premature graying of the hair, Nail dystrophy and pterygium (Panel B). Tongue was eroded, with marginal small white irregular patches (leukoplakia) (Panel C). These mucocutaneous signs were present since three years earlier. He had diffused fine crackles in both lungs on auscultation. Results from laboratory studies showed thrombocytopenia (platelet level, 35000 per deciliter) and high Erythrocyte Sedimentation Rate (ESR, 1st hour – 53 mm). He

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had severe restrictive pattern with TLC, RV, FEV1, FVC and FEV1/FVC equal to 3.12, 0.65, 1.86, 2.47 and 0.75, respectively (57%, 59%, 52%, 57% and 90% predicted, respectively). Computed tomography (CT) of his chest revealed inter lobular septal thickenings in both lungs' peripheral sites as 'honey combing' pattern which is consistent with pulmonary fibrosis (Panels D, E, F and G). These findings were suggestive of Dyskeratosis Congenita (DKC), which is a rare, progressive bone marrow failure syndrome characterized by mucocutaneous triad of reticulated skin hyperpigmentation, nail dystrophy, and oral leukoplakia [1]. He was given pharmacotherapy to alleviate his symptoms. We also started 1200 milligram of Perfenidon daily. At a 2-month follow-up, the patient's platelet level increased (74000 per deciliter) with androgen therapy, but there were no relief in his symptoms. Unfortunately, he expired one month later.

References

1. Jyonouchi S, Forbes L, Ruchelli E, Sullivan KE (2011) Dyskeratosis congenita: a combined immunodeficiency with broad clinical spectrum-a single-center pediatric experience. Pediatr Allergy Immunol 22: 313-319.