A rare case of acrogeria, Gottron type with borderline personality disorder

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Abstract

Acrogeria is a rare disorder that is characterized by premature aging of the distal extremities. It was first described by Gottron in 1941. Only about 50 cases have yet been reported worldwide. It is diagnosed clinically, and patients have a normal life expectancy. The disorder is seen from birth and could have an autosomal dominant or recessive inheritance. The classic features include a characteristic pinched face, thin lips, fine hair, skeletal defects, and thin, taut parchment-like skin of the extremities. We describe a case of Acrogeria, Gottron type, who also had a borderline personality disorder. However, there is very little information on the prevalence of psychopathology in patients having syndromes of premature aging.

Keywords: Acrogeria, borderline personality disorder, Gottron syndrome

Introduction

Acrogeria, Gottron type, is a rare non-progressive premature ageing syndrome characterized by accelerated aging of the distal extremities and other tissues without the involvement of internal organs and normal life expectancy. Both autosomal recessive and autosomal dominant inheritance have been reported with a higher incidence in females and are diagnosed clinically. There is no available literature on the prevalence of psychopathology in these patients. We therefore report a case of Acrogeria who presented with a Borderline Personality Disorder (BPD). BPD patients may first present to a family physician for behavioural issues like not listening, risk-taking, being emotionally reactive, and hence knowing about this would help physicians understand and deal with them more effectively.

Case Report

A 19-year-old female, first in birth order, born of a non-consanguineous marriage, was brought by her mother to the psychiatric outpatient department for behavioural problems over three years. Symptoms included repeated self-injurious behaviour, lack of interest in all activities, anger outbursts, irritability, fluctuations in mood, and excessive social media usage along with alcohol and cannabis consumption. Her academic performance also began to decline, and she got involved in multiple short-term relationships. She would have intense emotional fluctuations, anger outbursts, hostility towards teachers and family members, and multiple episodes of self-harming behaviour like consuming phenyl or excess of medicines when her demands were not met. As per the patient, she would feel empty and emotionally numb, and so she resorted to social media to make friends. She eventually started consuming alcohol and cannabis, dropped out of college in the 11th grade, and engaged in high-risk behaviour for thrill and novelty. She also expressed a strong desire to dye her hair a bright color, get multiple tattoos, visit haunted houses, and engage in other thrilling activities.
Her developmental history included normal milestones with average academic performance. Her mother had separated from her father when she was around three years old due to alcohol use and anti-social traits in him. Her relationship with her mother and younger brother was strained due to constant comparison between the two by the mother. She attained menarche at 13 years of age and had regular menstrual cycles.

Her physical examination revealed short stature for her age, with a height of 145 cm and a weight of 31.5 kg. She had sparse eyebrows; pigmentation over eyelids; dry, dull facial skin; pinched nose; mild prognathism with overcrowding of teeth and bleeding gums; sparse dry, thin hair on the scalp; lack of secondary sexual characteristics; lack of axillary hair; thin, dry, wrinkled skin with mottling over arms, hands, feet, and abdomen, with visible blood vessels; and rough ridges over the nails [Figure 1].

Her mother did realize that she was not growing up like the other girls of her age, but as she was working and the patient did not have any physical symptoms, she did not show her to any doctor. She was unaware of the patient’s skin condition. On mental status examination, the patient was well-kempt, cooperative, and communicative. She claimed her mood to be low, while her affect was stable and congruent to mood. She did not give any delusions, hallucinations, or suicidal ideations. Her judgement was preserved, and she had no insight into her illness. We admitted the patient for a thorough workup and took pediatric and dermatology opinions.

Psychological tests like the Rorschach Inkblot test were suggestive of a limited view of the world, rigidity, poor interpersonal functioning, and depressive tendencies; MMPI revealed the presence of emotional distress, impulsivity, rigidity, and she had average intellectual functioning with an IQ of 100 on Kamath Binet test.

On hormonal assays, FSH and LH were mildly raised. All routine blood investigations, chest X-ray, and ECG were within normal limits. Her skeletal survey revealed normal bone age, bone density, and alignment. Skin biopsy was suggestive of features of Acrogeria. It showed epidermal hyperplasia, increased thickness of the dermis, absence of eccrine and apocrine glands in the axilla.

Ophthalmological and ENT testing were within normal limits. DEXA scan revealed a high risk of osteoporosis with a T score hip of 4.3 and a T score lumbar spine of 2.5. Both pediatric and dermatology references confirmed the diagnosis of Acrogeria, Gottron type, but there was no treatment prescribed. Genetic testing could not be done as her mother was non-affording. The psychiatric diagnosis was BPD as per Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition,[8] for which the patient was started on Tab Valproate 200 mg twice a day, Tab Escitalopram 5 mg at night, and Tab Aripiprazole 5 mg in the morning. The patient has been following up in the OPD for the last six months, and is 70% better, though she still has impulsive behaviours, and so her Tab Escitalopram and Aripiprazole have been increased to 10 mg each.

## Discussion

Acrogeria, an extremely rare genetic syndrome with only about 50 cases reported,[12,4,5] involves mainly the distal part of the extremities and is characterized by a mild non-progressive form of skin atrophy giving it an aged appearance. It could be caused by mutations in the LMNA, ZMPSTE24, or COL3A1 genes.[10] The clinical features include thin, dry, wrinkled, and transparent skin over the extremities, easy bruising, nail atrophy or thickening, poikiloderma, and telangiectasia.[11,2] Our patient had bleeding gums and rough ridges over the nails. The characteristic facies of pinched face, hollow cheek, owl-eyed appearance, beaked nose, and thin lips[13] was not seen in our patient, although a pinched nose was present. A variety of skeletal defects have also been reported in such patients,[11] and her DEXA scans suggested a high risk for osteoporosis.

Our patient manifested most symptoms of BPD and concern over her thinning hair. Patients of Acrogeria or other premature ageing syndromes may perceive themselves to be less attractive due to the premature ageing process. This may cause or aggravate feelings of emptiness and low self-worth leading to other maladaptive behaviours. Patients with BPD have high rates of developing comorbid depression, anxiety spectrum disorder, substance use, and an eating disorder.[6,7] Our patient also had intense emotional fluctuations, multiple self-harm attempts, substance use, and feelings of loneliness. Anti-social personality traits and alcohol use in the father increased her vulnerability to developing a personality disorder. Separation of parents when she was young could have also shaped her emotions and behaviours. The need to engage in multiple superficial relationships, wanting to colour her hair bright could also be an effort to get validation of her appearance from others. Our

![Figure 1: Features of acrogeria](Image)
patient was bothered by her sparse hair as she could not style it. She would also use makeup routinely to make herself attractive. Researchers have not mentioned any body image disturbances in Acrogeria.

It will be interesting to study whether a perceived unattractive physical appearance increased her susceptibility for emotional disturbances or the genetic vulnerability led to her developing BPD. In view of the paucity of data, it remains important to explore the relationship between such premature ageing syndromes and low self-esteem, body image issues, and BPD.

Behavioural problems in adolescents are commonly seen, and it is necessary that a personality problem is not missed. Hair fall and skin problems may present first to the family physician. Though rare, premature aging can be picked up by only clinical examination. Creating awareness among all physicians is therefore important.

**Conclusion**

This patient presented to the psychiatrist for behavioural problems, and the general examination of the patient led to a suspicion of premature aging and diagnosis of Acrogeria. Such cases may approach the family physician as symptoms are not severe. The general examination is an important skill to be routinely practiced by every physician.

**Declaration of patient consent**

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

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**Conflicts of interest**

There are no conflicts of interest.

**References**

1. Acrogeria, Gottron Type. Genetic and Rare Diseases Information Center. Updated 2021 Nov 08. Available from: https://beta.rarediseases.info.nih.gov/diseases/6543/acrogeria-gottron-type. [Last accessed on 2022 Jan 01].
2. Burrows N. Genetic disorders of collagen, elastin and dermal matrix. In: Griffiths C, Barker J, Bleiker T, Chalmers R, Creamer D, editors. Rook’s Textbook of Dermatology. 9th ed. Hoboken, NJ: Wiley-Blackwell; 2016.
3. American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders: Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Arlington, VA: American Psychiatric Association; 2013.
4. Sanghi S, Grewal RS, Vasudevan B, Nagure A. A rare case of acrogeria. Med J Armed Forces India 2013;69:406‑8.
5. Marroofian R, Murdocca M, Rezaei-Delui H, Nekooei A, Mojarad M, Sangiulio F, et al. A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron type) in an extended consanguineous family. Clin Dysmorph 2018;27:88‑90.
6. Wayda-Zalewska M, Kostecka B, Kucharska K. Body image in borderline personality disorder: A systematic review of the emerging empirical literature. J Clin Med 2021;10:4264.
7. Shah R, Zaraini MC. Comorbidity of borderline personality disorder: Current status and future directions. Psychiatr Clin 2018;41:583‑93.