CASE REPORT
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Primary ovarian insufficiency in a xeroderma pigmentosum patient with consanguineous parents

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ABSTRACT

Introduction: Xeroderma pigmentosum (XP) is an inherited condition characterized by extreme sensitivity to ultraviolet rays from sunlight. It mostly affects the eyes and areas of skin exposed to the sun with some nervous system involvement. Diagnosis is done clinically. The prevalence of that catastrophic disease has been found to be at 1:1,000,00 in the United States and Europe. On the other hand, premature ovarian insufficiency was defined as the development of hypergonadotropic hypogonadism before the age of 40 years in women who have a normal karyotype. Case Report: A 30-year-old female who was a known case of xeroderma pigmentosum since birth. She came to the gynecology clinic complaining of oligomenorrhea for two years. Investigations were done and unfortunately this patient was discovered to have ovarian failure. There is a positive family history of consanguinity. Conclusion: Xeroderma pigmentosum is an autosomal recessive inherited disorder. For that reason, consanguinity is a very important risk factor. There is an increased risk of skin neoplasms and other internal neoplasms such as breast and uterine cancer. Moreover, premature ovarian failure is another rare disorder that we are presenting in this case in association with XP. A study that suggested a relation between XP and ovarian failure had found that three out of twenty XP patients study was found to have POF. In such cases, hormone replacement therapy will be danger as it increases the risk of breast cancer in those patients who are already at risk of internal neoplasms.

Keywords: Consanguineous parents, Premature ovarian insufficiency, Xeroderma pigmentosum

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INTRODUCTION

Xeroderma pigmentosum, commonly known as XP, is an inherited condition. It is inherited in an autosomal recessive manner and characterized by an extreme sensitivity to ultraviolet rays from sunlight. This condition mostly affects the eyes and areas of skin exposed to the sun [1]. Some affected individuals also have

There is a positive history of consanguinity between her parents. She does not smoke and no one of her family does. She studies psychology at the university.

When we examined her, Initials including: [height: 151 cm, weight: 77 kg and BMI: 33.7] and vitals including: [blood pressure 120/77, pulse 74 BPM brachial, temperature 37°C oral, respiratory rate 20 and O2 sat 100%]. Generally, the patient looks well, conscious and oriented to person place and time. The freckles are spread on the face, upper limbs, lower limbs, chest, and back. Scars from previous surgeries were noted on the nose, and multiple others were noted on the face. No special facial features are noted, no dysmorphism, no alteration was detected in the fingers and toes. Secondary sexual characteristics are well developed. Neck examination shows no palpable nodes or masses. The abdomen is soft and lax. Chest shows bilateral equal air entry with no added sound and the pericardium has shown normal first and second heart sounds with no obvious added sounds or murmurs. Neurological examination was done and it was unremarkable rather than that she mentioned progressive hearing loss which was not detected on examination. There was neither evident mental impairment nor cognitive deficit. Bone density showed osteopenia. Investigations including hormonal profile, TFT, blood grouping and vitamin D were given in Table 1, Table 2 and Table 3) and Female Pelvic Ultrasound (Figures 1–5).

The patient is on omeprazole 20 mg orally once daily and vitamin D. She is not following any measures of sun protection. Our plan was to give her hormone replacement therapy but because xeroderma pigmentosum patients have a high risk of internal cancers such as uterine and breast cancers the risk will increase more with the use of HRTs so the role will be by treating the symptoms of premature ovarian inefficiency. On the other hand, she requires a condensed education regarding her issue as she had multiple occasions of skin cancer that were treated with skin excision.

Table 1: Hormonal profile: Taken day-3 post last menstrual cycle

Test	12/3/2015	13/5/2014	Reference range
FSH	47.2 pmol/L	50.6 pmol/L	
LH	31.1 mIU/L	29.1 mIU/L	
Prolactin	399.6 mIU/L	167 mIU/L	
GH	0.05	----------	
Estradiol	92 pmol/L	49 pmol/L	26–125 pmol/L
Progesterone	0.969	0.580	83–622 nmol/L

Table 2: Thyroid function test (TFT)

TFT	28/3/15	12/3/15	23/10/14	1/9/14	13/5/14	24/4/14	21/11/13	15/8/13	13/3/13	Reference range
TSH	0.381	0.591	2.49	3.47	0.628	1.191	3.43	4.88	1.81	0.27–4.2 uIU/L
T3	3.9	4.4	4.3	3.7	4.6	4.34	3.63	4.13	3.64	2.8–7 Pmol/L
T4	18.1	19.6	17.0	16.8	17.5	18.77	13.00	19.27	15.23	12–22 Pmil/L

DISCUSSION

Xeroderma pigmentosum (XP) is known to be an autosomal recessive inherited disorder. One other rare subtype of the disease that is called XP dominant type is inherited as an autosomal dominant disorder [16]. For that reason consanguinity in the parents of the patients of XP has been shown to be an important etiologic factor [17]. According to a study, it has been reported to be 92.8% in XP patients in Libya [18]. Studies from Egypt, Pakistan, and Nigeria have shown a high incidence of the disease as these countries are considered to be among the countries, which has high rates of consanguinity [19, 20]. On the contrary, a series of four cases of XP in which consanguinity had not been observed in the parents of those patients have been described [21]. In another case, the patient was born to consanguineous parents. However, there was a varying degree of the disease progression and severity even among sibling who are affected [17]. The patient in the present case also was born to parents who had a history of consanguineous marriage and with variable degrees of affection and progression among the siblings. Her two brothers who are affected with the same disease showed slower progression of the disease with less affection as they never had a history of skin cancer and the pigmentary changes were not involving the same amount of their body as our patient has. Although the disease affect more commonly patients who are the product of consanguineous parents. Stephanic Christen-Zaech et al. reported a case that showed that XP has occurred in an uncle and nephew [22]. In XP, the main defect is in the nucleotide excision repair (NER), in which this leads to deficient repair of DNA damaged by ultraviolet radiation [23]. Nine gene mutations (namely: XPA, ERCC3, XPC, ERCC2, DDB2, ERCC4, ERCC5, ERCC1, and POLH) had been reported. As a result, the disease had been classified by complementation groups (XP-A, XP-B, XP-C, XP-D, XP-E, XP-F, XP-F, XP-G). Researches have also described another form of the disease that is called XP variant (XP-V). As with the other subtypes of XP the symptoms and signs of the other subtypes may also be seen in XP-V patients. As a result, the preferred method of laboratory diagnosis is functional testing to screen cells for abnormalities in DNA repair. Simply