# Xeroderma Pigmentosum: A Case Report

**Sam Park, DDS  Murray Dock, DDS, MSD**

*Dr. Park is in private practice, Chicago, Ill; Dr. Dock is associate professor of clinical pediatrics and residency program director, Division of Pediatric Dentistry, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio.*
*Correspond with Dr. Dock at murray.dock@chmcc.org*

## Abstract

This paper presents a case study of a child with xeroderma pigmentosum (XP). The disease results in sensitivity to UV radiation as a result of reduced activity in a defective enzyme responsible for DNA repair. Affected individuals have a variety of clinical symptoms, which may include problems of the skin and oral mucosa, ocular manifestations, and neurologic impairment. A number of precautions must be taken when treating these patients, which include proper shielding from damaging light and the selection of suitable dental materials. The necessary measures required when treating patients with XP are reviewed in this report. (*Pediatr Dent*. 2003:25:397-400)

**KEYWORDS: XERODERMA PIGMENTOSUM, UVA RADIATION, UVB RADIATION, CURING LIGHTS**

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Xeroderma pigmentosum (XP) is a rare, autosomal recessive disease characterized by severe sensitivity to all sources of ultraviolet (UV) radiation and occurs in approximately 1 in every 250,000 live births.[1,2] The disease usually manifests early in life after the child's first exposure to sunlight. A severe sunburn after a short exposure to the sun that lasts much longer than expected is characteristic.

Light is divided into 3 categories based upon wavelength: (1) ultraviolet radiation (<400 nm); (2) visible light (400-700 nm); and (3) infrared or heat (>700 nm). In turn, ultraviolet radiation is further divided into UVA (320-400 nm), UVB (290-320 nm), and UVC (200-290 nm). UVC rays are usually absorbed by the atmosphere and ozone and do not reach the earth's surface. UVA and UVB rays, however, are both capable of penetrating the atmosphere.[3] It is the UVA and UVB wavelengths of light that are of concern in the XP population.

**Table 1. Skin Lesions**

| |
|---|
| Hyperpigmented macules |
| Solar lentigines |
| Melanomas |
| Squamous cell carcinoma |
| Basal cell carcinoma |
| Actinic keratosis |
| Telangiectasia |

The condition is definitively diagnosed by measuring the DNA repair defect from skin or blood obtained from the patient.[4] The abnormality is in the nucleotide excision repair mechanism which is responsible for recognizing and repairing UV light damaged regions of DNA. There are 8 XP repair genes which have been identified: XPA through XPG and 1 variant type. Phenotype presentation relates to the degree of reduced activity in the defective enzyme responsible for DNA repair.[5]

There are a variety of of physical findings characteristic of the disease. Involvement of the skin and mucosa, eyes, and central nervous system are the most common (Tables 1, 2, and 3). The lesions most pertinent to the dental practitioner are those found in the head and neck region. This is where 65% of the melanomas and 97% of the basal and squamous cell carcinomas arise.[6,7] Oral manifestations usually involve the lips, anterior portion of the tongue, and buccal mucosa. The most common lesions include squamous cell carcinomas of the lips and tongue, cheilitis of the lips, glossal telangiectasias, and leukoplakia.[8,9]

**Table 2. Ocular Lesions**

| |
|---|
| Conjunctivitis |
| Ocular squamous cell carcinoma |
| Blepharitis |
| Symblepharon |
| Keratitis |
| Corneal opacities |
| Tumors of the eyelids |
| Blindness |

**Table 3. Neurological Manifestations**

| |
|---|
| Microcephaly |
| Mental retardation |
| Craniosynostosis |
| Choreoathetosis |
| Cerebellar ataxia |
| Sensorineural deafness |

Currently, there is no known cure for XP. The DNA damage that is suffered is cumulative and

Figure 1. Physical protection.

eventually leads to malignant growths. Prevention and early treatment can limit the problems that arise. The purpose of this article is to inform the dental practitioner of the precautions necessary when treating these patients.

## Case report

A 10-year-old boy with a history of XP was referred to the dental clinic at Cincinnati Children's Hospital Medical Center (CCHMC) by his pediatrician with a chief complaint of pain from an abscessed tooth. The pain started several days prior and his physician at that time prescribed a course of clindamycin. His medical history included reactive airway disease exacerbated by stress, with symptoms occurring approximately once a month. His surgeries included 2 sets of ear tubes, over 30 excisions of basal cell carcinomas and scleromas from various parts of his body, and 1 prior dental rehabilitation under general anesthesia. His lesions included numerous areas of hyperpigmented macules and papules with several areas of telangiectasia. Other lesions were basal cell carcinomas found on the vertex of the scalp, left parietal scalp, left mandible, right and left forearms, right nasal sidewall, right cheek, left medial chin, and left lateral chin. His medications included hydroxyzine, prednisolone acetate eye drops, albuterol inhaler, and over-the-counter lotions and sun block. His last dental visit was 5 years ago.

Prior to his dental appointment, a thorough evaluation of all light sources in the clinic was made with the assistance of the clinical engineering staff of the hospital. Light switches which control the overhead lights and the dental lamp in the operatory were bolted in the off position. Hallway lights were dismantled to minimize light from entering the room upon opening and closing of the door to the operatory. The window in the door was sealed to further decrease the chance of light exposure.

The patient arrived at the hospital wearing a full-body protective covering consisting of an all-black helmet, ski gloves, ski jacket, and double woven jeans to filter all UV light (Figure 1). He was met at the hospital entrance with a wheelchair to allow for more rapid transport to the dental clinic. Once in the dental operatory, with all of the lights turned off, the suit was removed and he was partially covered with a blanket.

The oral examination was conducted in a completely darkened room using only an incandescent bulb in a flashlight to illuminate the mouth. Incandescent light does not emit harmful UV wavelengths. The patient was in the

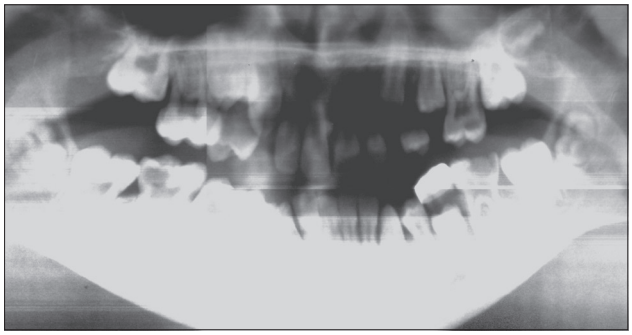

Figure 2. Panelipse film.


Figure 3. UVA and UVB radiometers (National Biologic).

mixed dentition with a stainless steel crown on his mandibular left second primary molar and no other restorations. All 4 of his permanent first molars had decay with pulpal involvement affecting the mandibular molars (No. 19 and 30). Bite-wing and a panelipse film (Figure 2) were made which revealed a periapical radiolucency associated with the mandibular left first molar (No. 19). The maxillary left first molar (No. 14) had a large lesion that did not invade the pulp, and, upon clinical examination, the maxillary right first molar (No. 3) was found to have incipient decay. There were no other carious teeth identified. His oral hygiene was poor with moderate generalized gingivitis. All other soft tissues were normal. His overbite was 50% with a 2 mm overjet with severe maxillary and mandibular crowding. His skeletal profile was convex.

All treatment options, along with advantages and disadvantages of each, were discussed with the mother. A treatment plan was agreed upon, which included extracting the cariously involved molars rather than subjecting the patient to multiple appointments of endodontic and restorative procedures. Due to the extent of his needs and the complexity in performing the procedures in the clinic, treatment was scheduled in the operating room under general anesthesia.

The patient had been to CCHMC's operating room multiple times, so the hospital staff was very familiar with his needs and all precautionary measures were taken to ensure his safety. The preoperative holding area and postanesthesia care unit both prepared a specially darkened room for his arrival. The operating room where the procedure

was performed was also modified. All windows to the operating room were sealed and a sign was placed on the door prohibiting entrance once the patient was transported to the room. UVA and UVB light meters were used to measure the wavelength of all light sources to which the patient was exposed (Figure 3). The lights throughout the hallway to the operating room, the overhead lights as well as the operating lights and dental lamp in the operating room were all found to be negative for emitting UVA/UVB wavelengths. The dental curing light (Dentsply QHL75), however, did not register within satisfactory limits. The reading from the authors' UVA/UVB meter registered 1.3 $mW/cm^2$ for UVA and 0.03 $mW/cm^2$ for UVB.

Treatment was successfully completed under general anesthesia. All permanent first molars were extracted without complication, hemostasis was achieved, and Gelfoam and 1, 3-0 chromic gut suture was placed in each extraction site. The mandibular left second primary molar was also extracted due to its imminent exfolation, and adequate hemostasis was achieved with gauze pressure without sutures. A rubber cup prophylaxis was completed at the end of the treatment. The child was placed on a strict prevention protocol which included meticulous oral hygiene, dietary counseling, recommendation of the use of an over-the-counter fluoride rinse, and routine recalls.

## Discussion

There are a number of important issues that must be considered when providing dental treatment for patients with XP. Of utmost importance is adequate office preparation prior to the patient's arrival. A UV light meter should be used to test any light source that the patient may be exposed to during the visit. These include overhead lights, dental lamps, viewboxes, fiberoptic lights, computer screens, and dental curing lights. While most of these light sources will read "zero" for UVA and UVB radiation, it is important that all light sources be tested prior to patient exposure. Radiation emitted by radiographic equipment falls outside the range that is harmful to patients with XP and is not contraindicated. All windows should be properly sealed to prevent any sunlight from entering into the office. A proper seal on a window can usually be accomplished by simply drawing the blinds. Evening or night appointments will further limit the risk of exposure to sunlight. A visit by the parent or guardian to the office prior to the patient's appointment would be helpful. Most, if not all, parents will have access to a UV light meter. All necessary precautionary measures can be reviewed at this time.

A dilemma that may occur during treatment of a child with XP is the inability to use traditional curing lights. The range at which most composite restorative material is optimally cured is between 450 and 470 nm. The majority of curing lights are equipped with a filter that allows light at only 450 nm to be emitted; however, despite this, many of these units emit light in the UVA/UVB range. The problem relates to filters being ineffective or wearing out over time. Thus, the use of sealants and composite restorations may be problematic. Light emission diode (LED) curing lights are manufactured to emit light at a wavelength at only 450 nm, which seems like a reasonable solution to the problem. Testing of the authors' LED curing unit (Caulk NRG) however, registered a UVB reading of 0.01 $nm/cm^2$ and a UVA reading of 0 $nm/cm^2$. Plasma arc curing (PAC) lights emit UV wavelengths in a range between 380 to 495 nm which contraindicates the use of these lights. Other alternative methods of curing materials include the use of argon lasers. This device produces light over a narrow band of wavelength, 457.9 to 514.5 nm and consequently could be used in patients with XP.[10]

All light sources, regardless of the manufacturers' claims, should be tested with calibrated light meters prior to use. Any reading above 0 $nm/cm^2$ should contraindicate the use of that unit. Autopolymerizing sealants and other restorative materials such as glass ionomers are a good alternative when a suitable curing unit is unavailable.

Patients with XP often do not maintain routine dental appointments due to the difficulty in arranging for care. Therefore, they are at increased risk for dental disease. Consequently, it is important that the dental community be educated about this disease and not be hesitant to provide treatment for these patients. Meticulous oral hygiene including proper brushing and flossing and fluoride rinses, routine recall appointments, and diet counseling are imperative. Additionally, due to the increased risk of oral lesions, careful examination of soft tissues should be regularly completed. Patients should be encouraged to apply sunscreen liberally to the lips to help reduce the risk of lesions to this area. A list of considerations when seeing a child with this disease is outlined in Table 4. Medical colleagues should be encouraged to emphasize the importance of regular dental care to this population of patients. With the precautions outlined above, providing dental care to XP sufferers can be very rewarding to patients, their families, and practitioners as well.

**Table 4. Considerations When Caring for a Child With XP**

| |
|---|
| Test all light sources in office |
| Dental lights |
| Viewboxes |
| Fiberoptic lights |
| Curing lights |
| Computer screens |
| Seal windows (closing blinds should be adequate) |
| Evening appointments preferable |
| Institute home preventive program |
| Emphasize routine recalls |
| Careful examination of soft tissues for lesions |
| Use of appropriate composite/resin materials and curing methods |

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## Abstract of the Scientific Literature

### Cephalometric Comparison Between the Pendulum and Herbst Appliances

Class II skeletal malocclusion can be due primarily to mandibular retrusion or maxillary protrusion. When treatment planning, the Herbst appliance has more often been used to treat patients with mandibular retrusion, and the pendulum has been used for patients with maxillary protrusion. It was the purpose of this study to compare the effects on Class II malocclusion of the Herbst appliance followed by fixed appliance and the pendulum appliance followed by fixed appliance to determine any morphological differences achieved. This retrospective study evaluated skeletal Class II patients treated with: group 1=stainless steel crown Herbst (N=30); group 2=acrylic-splint Herbst (N=30); or group 3=pendulum (N=30). All subjects were matched for age/sex with a mean of 12 years and a girls:boys ratio of 2:1. After cephalometric analysis was done at initiation of treatment and before fixed appliance therapy was started, the results showed no major differences between groups at the start of treatment. After treatment was completed, for both Herbst appliances the mandible grew approximately 4.5 mm (no data is given for the pendulum group). Greater maxillary molar distalization was found with the pendulum appliance. The authors conclude that the Herbst appliance achieves results by 50% tooth movement and 50% skeletal changes, whereas the pendulum appliance has no skeletal component.

**Comments**: The article presents data to support the use of a functional appliance to advance the mandible in cases of retrusion. Unfortunately, no long-term follow-up data was given. This would have been very useful, since most of the controversy with the use of functional appliances lies in the long-term stability and not the short-term results. **KV**


*Address correspondence to James A. McNamara, Department of Orthodontics and Pediatric Dentistry, School of Dentistry, University of Michigan, Ann Arbor, MI 48109-1078.*



**Burkhardt DR, McNamara JA, Baccetti T. Maxillary molar distalization or mandibular enhancement: a cephalometric comparison of comprehensive orthodontic treatment including the pendulum and the Herbst appliances. *Am J Orthod Dentofacial Orthop.* 2003;123:108-116.**

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