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Mini review

Xeroderma pigmentosum family support group: Helping families and promoting clinical initiatives

Michele Milota[a], Debby Livingstone Jones[a], James Cleaver[a,b], Ijaz S. Jamall[a,*]

[a] XP Family Support Group, 8495 Folsom Blvd #1, Sacramento, CA 95826, USA
[b] Departments of Dermatology and Pharmaceutical Chemistry, Helen Diller Family Comprehensive Cancer Center, University of California San Francisco School of Medicine, Box 0808, UCSF, San Francisco, CA 94143-0808, USA

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ABSTRACT

The past two decades of research into Xeroderma pigmentosum (XP), an autosomal recessive disease, has been marked by significant progress in understanding the molecular basis of this rare disease. More importantly, especially from the perspective of the affected families, is that this knowledge has been applied to diagnose the condition both *in utero* as well as in the very early days of life. The eight known XP genes and their different phenotypes present a number of challenges that the XP Workshop sponsored by the NIH in 2010 has highlighted. There is little current treatment specifically designed for any of the XP types other than standard dermatological and neurological evaluations and care. The Xeroderma Pigmentosum Family Support Group (XPFSG) is dedicated to serving families with children and adults with all forms of XP and to help them better understand the condition, to identify practical measures which can be taken by the XP patients and their families to mitigate the effects of the disease, and to serve as patient advocates to help families discuss issues with their physicians. We summarize our efforts in terms of outreach within the US and abroad to affected families and discuss XPFSG-sponsored clinical initiatives that include molecular diagnoses, treatment, and initial proof-of-concept studies that can, if successful, improve the lives of XP patients in the near term.

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1. Introduction

1.1. The XPFSG—what it is and what it does

"Every child deserves a day in the sun". This is the motto for a small grass roots organization founded by Michele Milota from Sacramento, California in December 2005. XP, a rare, autosomal defect in the DNA repair system, is responsible for numerous clin-

* Corresponding author at: Director of Research-XPFSG and Risk-Based Decisions, Inc., 1540 River Park Drive, Suite 203, Sacramento, CA 95815, USA. Tel.: +1 916 923 0570.
E-mail address: ijamall@xpresearch.org (I.S. Jamall).

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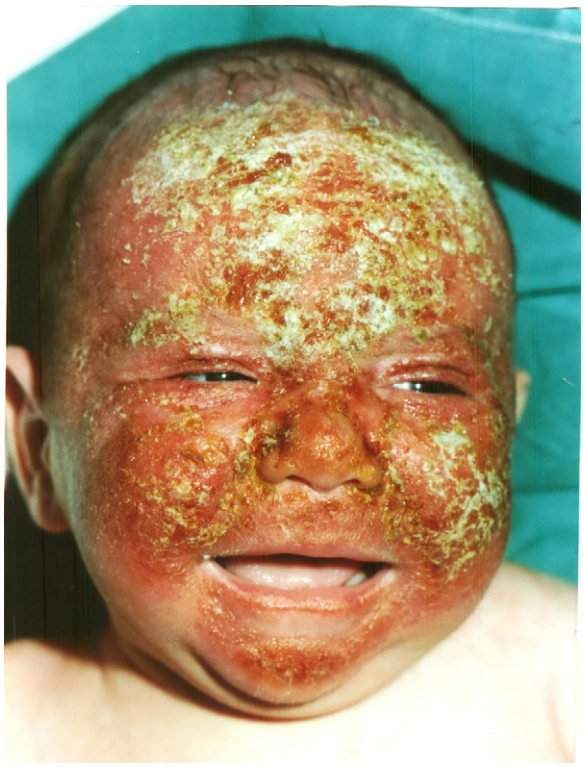

Fig. 1. Infant with XPA showing extensive skin lesions commonly associated with disease.

ical problems for patients and their families. XP prevents those afflicted by the most common forms of the disease from engaging in any activity in the sun or other sources of ultraviolet radiation (UVR). Michele, along with other families who share in the daily tribulations of this disorder, have established themselves as a round-the-clock information-friendly group of compassionate soldiers fighting for the lives of their own family members and others affected by XP.

The goal of the XPFSG is to help all individuals affected by this genetic condition. Due to the normal inclinations of all individuals to live within the socially and biologically mandated rhythms of natural light, it is a formidable task to keep XP kids from being exposed to the harmful effects of the sun (Fig. 1). This small group of highly motivated individuals is dedicated to improving the quality of life for those who suffer from XP in the form of emotional support, education, assisting that the correct diagnosis is made, effective disease management options, and ultimately, a cure. The vast number of DNA test models, charts and grim cellular findings might make the latter seem unlikely but this Group will hear none of that. "We look for a cure. . .always. . .but, if it doesn't come, it won't be because we didn't give it every ounce of effort that is within us" touts Michele, the mother of a seven-year old daughter with XPC.

1.2. XPFSG and fundraising

From a fundraising perspective, XP ranks low on the numerical scale with organizations that support better known diseases like Breast Cancer, Leukemia, Muscular Dystrophy, and other such causes that regularly make the headlines. XP affects about one person in every 1.5 million people in the United States. Fundraising, and an unyielding dose of tenacity, is the primary vehicle for success for the XPFSG. Everyone affiliated with this Group is encouraged (and assisted) to engage in fundraising. For most non-profit groups, fundraising is a universally understood and accepted way to gather the money a 'giving' people wish to share. . .as a means to an end.

Table 1
XP individuals affiliated with the XFSG by gender, XP type and current status.

Gender	Age (as of 2010)	XP Type	Status
M	8	Unknown	Alive
F	8	C	Alive
F	15	D	Alive
F	23	A	Deceased
F	10	A	Alive
F	8	C	Alive
M	22	C	Alive
M	13	Unknown	Alive
F	7	C	Alive
F	25	G	Alive
F	12	C	Alive
M	7	C	Alive
M	16	C	Alive
F	14	C	Alive
M	NA	C	Alive
M	16	C	Alive
F	25	A	Alive
F	23	A	Alive
M	7	C	Alive
M	55	V	Alive
F	55	E	Alive
M	17	C	Alive
F	26	C	Alive
M	35	V	Alive
M	4	C	Alive
F	2	D	Alive
F	10	D	Alive
F	8	C	Alive
M	NA	C	Alive
M	NA	C	Alive
F	18	C	Alive
M	11	A	Deceased
F	16	A	Deceased
M	33	V	Alive
F	29	V	Alive
F	8	C	Alive
M	4	A	Alive
F	17	C	Alive
F	18	Unknown	Aive
M	16	C	Alive
M	16	Unknown	Alive
F	15	Unknown	Alive
F	17	D	Alive
F	16	C	Alive
F	11	C	Alive
M	5	C	Alive
F	5	C	Alive
M	8	D	Alive
F	18	Unknown	Alive
F	15	Unknown	Alive

For this Group, it is an attempt to gain recognition from a 'largely unaware' audience. These 'mostly' children with XP suffer from a variety of medical problems ranging from multiple skin cancers to neurological deficiencies and a shortened lifespan, depending on the severity of their disease and the quality of their care. Some individuals live longer, but the average XP child lives no more than three decades (median age 29 with neurodegeneration; 37 without. See Bradford P. NCI). In the past 16 months, the XPFSG has lost four XP patients, ranging in age from 15 to 32 years of age (Table 1).

1.3. XPFSG and family support

As rare as XP is, the presence of various genotypes and the challenges met by each patient ranges in appearance and severity. For all of those diagnosed with XP, there is one hurdle that is apparent from the time a child begins to explore the world outside the home. From as early as three or four years old, XP children begin to realize their unique challenges in social settings. Everything they see on television or through their UV-tinted windows is 'fantasy' for