

The New Face of Genetics

In the early 1980's an important educational documentary called "The Pearls of Dysmorphology" was made by Dr. Bryan Hall of the Greenwood Genetics Center. This film documented the dysmorphic features of some 67 different inherited genetic conditions and was created as a tool for medical professionals to learn the specific morphology of individuals affected with these conditions. With limited technology, Dr. Hall filmed his patients in a clinical setting pointing out the hallmark characteristics, or "pearls", of each condition. While the film still holds substantial educational value, the medical community is in need of an updated version of this important tool -- a film that not only incorporates the knowledge we have gained about these genetic conditions during the last 30 years, but also reflects a commitment to treating individuals with these conditions as human beings rather than specimens. Our hope is that this film will give future genetic counselors and physicians a more robust understanding of rare genetic conditions and also model an attitude of respect for the humanity of future patients.

In our film, "The New Face of Genetics", we will present individuals with genetic conditions who will introduce themselves to viewers and then demonstrate hallmark characteristics of his or her condition. This provides students the opportunity to study a genetic disorder on a live human being rather than from still photographs in a textbook. Not all aspects of a condition can be captured in still photography. For example, many individuals with Marfan syndrome will have a displaced lens in the eye. This can be detected by a characteristic movement of the lens as it is displaced upward. Most importantly, we believe that the format we are proposing will afford us the opportunity to present these individuals in a manner which also captures their humanity and beauty. Viewers will get to see our participants talk, move, or smile, making this a departure from the "patient-as-a-specimen" model that most educational medical literature utilizes.

We intend to make a pilot of this new film by creating profiles for three to five genetic conditions. This would act as a template for future graduate students to add to the project eventually creating a compendium of inherited genetic conditions with dysmorphic features. This entire process would happen through a collaboration with Rick Guidotti of Positive Exposure, an arts organization dedicated to working with individuals living with genetic difference. The script of the film will reflect the most current information on the conditions as well as input from well-regarded geneticists. We also will develop partnerships with the relevant support organizations for these diseases. They will be instrumental in our goal to present the most pertinent, up-to-date information about a disease, and they will also help us identify potential participants in the film. Finally, Mr. Guidotti's extensive photography and videography experience will be essential to ensuring that the film is visually appealing by current media standards.

Our hope is that this film will eventually be used by medical students and professionals as a tool to learn all of the distinct characteristics of specific genetic conditions. But more importantly, we hope to highlight the humanity of these individuals alongside the key medical information.

*Figure 1: An Individual with Marfan Syndrome in the Pearls of Dysmorphology Video
(Hall 1998)*



*Figure 2: An Individual with Marfan Syndrome in a Positive Exposure Photograph (Rick
Guidotti 2008)*



Figure 3: An Individual with Noonan Syndrome in the Pearls of Dysmorphology Video

(Hall 1998)



Figure 4: An Individual with Noonan Syndrome in a Positive Exposure Photograph (Rick

Guidotti 2008)



