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## Effect of Photographs of Visible Genetic Conditions on Quality of Life Perceptions

Christina Falugi

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EFFECT OF PHOTOGRAPHS OF VISIBLE GENETIC CONDITIONS ON QUALITY OF LIFE  
PERCEPTIONS

by

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EFFECT OF PHOTOGRAPHS OF VISIBLE GENETIC CONDITIONS ON QUALITY OF LIFE  
PERCEPTIONS

A  
THESIS

Presented to the Faculty of  
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and  
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MD Anderson Cancer Center  
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of the Requirements

for the Degree of

MASTER OF SCIENCE

by

Christina Jessica Falugi, BS  
Houston, Texas

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# EFFECT OF PHOTOGRAPHS OF VISIBLE GENETIC CONDITIONS ON QUALITY OF LIFE PERCEPTIONS

Christina Jessica Falugi, BS

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Historically, medical photographs are used to demonstrate dysmorphic features and characteristic presentations of genetic conditions. Traditional, pictorial depictions of genetic conditions typically involve nude subjects against walls to emphasize their features. These stark, black and white photographs may negatively influence students' perceptions of the depicted individual. Natural photographs which include individuals in non-clinical environments may impact perceptions. To assess the influence of photographs on a viewer's perception, 649 students from medical, nursing, genetic counseling and dental programs were surveyed in a cross-over study. Students were randomized to view a traditional or a natural photograph of three distinct genetic conditions followed by a natural or a traditional photograph, respectively, of the same conditions. Perceptions of the individual and their quality of life were assessed using Likert scale and yes/no adjective-association questions. Affected individuals were more often associated with positive characteristics (e.g. beautiful, respectful, intelligence, higher quality of life, etc.) when presented in natural settings and negative characteristics (e.g. degrading, institutionalized, humiliating, neglected, etc.) when depicted in traditional photographs ( $p < 0.05$ ). These associations were evident regardless of which photograph was viewed first. Furthermore, the nature of the first photograph influenced how much more positive or negative the second photograph was perceived. Although overall trends remained the same, the type of healthcare program influenced the magnitude of the effect; genetic counseling students tended to have more positive perceptions than medical, dental, and nursing students ( $p < 0.05$ ). These results suggest that using natural images for healthcare education may positively impact viewer's perceptions and potentially neutralize negative biases associated with genetic conditions.

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**ABBREVIATIONS**

QoLA- Quality of Life Assessment

PA- Perception Assessment

CdL- Cornelia de Lange syndrome

## BACKGROUND

Photographs have various roles in modern life. They are used to capture sentiments, solidify memories, and evoke emotions from every day moments and significant life events. They also lead to judgments based on visual impressions from the photographs, particularly when the pictures include a person. The portrayal of subjects within photographs can influence the viewer's perception of the individuals' traits (Dollinger, 2002). For instance, photographic subjects who are smiling or pictured with others are perceived to be more physically attractive than isolated or unsmiling subjects (Dollinger, 2002). Photographs are utilized as instructive tools in the education of healthcare providers, and therefore, it is possible that these photos may influence providers' perceptions as well.

Photographs in healthcare textbooks are useful in illustrating visually-evident features of certain conditions and allow for accurate diagnoses based on characteristic presentations. Historically, medical photographs as commonly presented in textbooks and other literature (hereafter referred to as "traditional" photographs) are black and white, stark, and unflattering to their subjects, portraying them nude or partially and recording their vulnerabilities (Jones, 1996). These pictures are largely alienating, humiliating, and dehumanizing towards patients (Jones, 1996). Despite the unfavorable depiction of patients, these pictures are widely utilized for their instructive value based on the emphasis on the subjects' features. This is especially relevant when describing genetic disorders, as individuals with genetic conditions can exhibit facial and/or physical differences that separate them from their peers. Impressions made from these images can lead to consequences. Due to their distinct appearance, these patients can suffer from stigmatization (Ablon, 2002). Both perceived and enacted stigmatizations have been associated with negative life experiences, particularly social interactions and desirability (Strauss, 2007). Studies have demonstrated that there is a significant difference in an individual's perceived personality based on their attractiveness (Serketich, 1997 and Dumas, 2001). For example, when adults viewed photographs of children, those that were perceived as attractive were associated with social competence while children who were perceived to be unattractive were judged to be more aggressive and anxious (Serketich, 1997). Furthermore, children who were viewed as attractive were treated more

positively and exhibited more positive behaviors than those who were judged as less attractive (Langlois, 2000).

Similar observations have also been made between patients and their medical providers, where a patient's appearance can influence their provider's opinion of them and the relationship between the two. Huizinga et al. (2009) found that physicians' respect for patients decreased as the patients' BMI increased. An additional study also revealed that patients with a higher BMI were perceived to be less adherent to medications by their physicians (Huizinga, 2012). These examples demonstrate how patients' appearances and the stigma associated with obesity affected the judgements made by physicians during their encounters.

A not-for-profit organization working towards changing these experiences and impressions is *Positive Exposure*© (positiveexposure.org). Founded by Rick Guidotti, *Positive Exposure*© “utilizes photography and video to transform public perceptions of people living with genetic, physical and behavioral differences” (Guidotti, 2012). The photographs taken by *Positive Exposure*© are portraits that display subjects in a non-clinical environment. These kinds of photographs (hereafter referred to as “natural” photographs) are different from the traditional photos by demonstrating the physical features of the subject in a real life setting. *Positive Exposure*© uses media to challenge social prejudices associated with genetic conditions and empower both individuals captured within the images and their viewers (Sutton, 2006). Individuals who participate in *Positive Exposure*© photoshoots report increased self-esteem and develop newfound self-perceptions, including increased attractiveness and confidence when comparing how they felt before and after completing a photoshoot (Sutton, 2006). This difference in mentality could have important implications for individuals viewing these natural photographs and their perception of individuals with genetic conditions.

Previous unpublished research demonstrated that there is no difference in students' learning regardless of whether a traditional or a natural photograph of a visible genetic condition is viewed (Wood, 2013). In this study, 156 genetic counseling students, 150 medical students, and several other student groups were shown one version of a photograph, traditional or natural, and asked a series of

didactic learning questions. While learning remains unaffected, data from this study and *Positive Exposure*© suggests these natural photographs could more positively impact quality of life perceptions of individuals with genetic differences and combat associated stigma within the medical community. Quality of life is the perception of an individual's position in life socially, culturally and environmentally (Phillips, 2006). This idea includes an individual's physical health, psychological state, independence, social and environmental relationships, and personal beliefs (Phillips, 2006). Wood *et al.* (2013) also examined whether there was a difference in quality of life perceptions between traditional and natural photographs when viewed by students from various health care programs. The results revealed that students who viewed the natural photographs rated the individuals depicted more positively ( $p < 0.05$ ).

Further research is needed to examine the differences in perception between traditional and natural photographs. Examining students from a variety of educational programs would provide important information about their perceptions, particularly when they are able to assess both the traditional and natural images when considering quality of life and descriptive adjectives. Therefore, this study aimed to determine if a student's perception of the depicted individual's quality of life varies by the type of photo presented, order of the photograph viewed, and educational background of the learner.

## METHODS

### *Study Participants*

The study population included genetic counseling, medical, nursing, and dental students. Eligible participants included genetic counseling students from the 35 North American genetic counseling programs, medical students from the University of Texas Medical School at Houston, University of Texas Medical Branch at Galveston, Baylor College of Medicine, and Virginia Commonwealth University, nursing students from the UT Health School of Nursing, and dental students from the University of Texas School of Dentistry at Houston and Virginia Commonwealth University. This study was approved by the institutional review boards at the University of Texas Health Science Center (IRB #HSC-MS-15-0503) and Baylor College of Medicine (IRB #H-37609). Virginia Commonwealth University IRB approval was not necessary as no study collaborators were at that institution. Healthcare students at these institutions received an email inviting them to participate in the survey between October 30, 2015 and February 29, 2016. The questionnaire was available to each group of students at different times within this period. The email invitation was sent to genetic counseling program directors and faculty at the other institutions to disseminate to their respective students. This email included a brief introduction to the study and a link to the online survey. Clicking the survey link served as informed consent in this study.

### *Study Design*

A cross-over study design was used for this project. Participants were randomized into two groups, arm A and arm B, based on whether they were born on an even or odd numbered day, respectively. Each of the study participants viewed all 6 images, with the only difference between the two arms being the sequence the images were presented. In the first part of the study (period 1), the students in Arm A viewed the traditional photographs. This was followed by the second part of the study (period 2) in which these same students viewed the natural photographs. Conversely, students in Arm B viewed the natural photographs in period 1 and traditional photographs in period 2.

### *Study Participant Recruitment and Measures*

Eligible students received an email invitation (Appendix A) which included a link to the questionnaire (Appendix B). Participants were surveyed through REDCap, an anonymous online data collection system. Three genetic conditions, Marfan syndrome, Cornelia de Lange syndrome, and Noonan syndrome, were depicted in both the traditional and natural formats (Figure 1). The natural photographs used within this study were all *Positive Exposure*© images that were presented in black-and-white, similar to the traditional photos. Each photograph was followed by questions related to that photograph/condition.



Figure 1: Natural Photographs Assessed in this Study

A. Marfan syndrome B. Cornelia de Lange syndrome C. Noonan syndrome  
Traditional Marfan syndrome image not included as permission was not granted. Traditional Cornelia de Lange and Noonan syndrome photographs are not included as permission for usage was not granted by Elsevier B.V. These pictures are image C on page 90 and image A on page 123 of the 5<sup>th</sup> Edition of Smith's Recognizable Patterns of Human Malformation.

The survey consisted of demographic questions and questions designed to assess students' perception of the individual within each photograph. The first portion included a Quality of Life Assessment (QoLA) which used a 5-point Likert scale ranging from least representative (1) to most representative (5) of that trait to allow students to rate each photographic subject in terms of the following 7 characteristics: attractiveness, popularity, competence, intelligence, hardworking, communicativeness, and quality of life. The second part consisted of a Perception Assessment (PA) where students were presented with a list of 18 adjectives and indicated whether or not the corresponding

photograph brought each of them to mind. Positive adjectives included creative, respectful, beautiful, individualistic, flattering, artistic and relatable. Negative adjectives included degrading, humiliating, neglected, depressing, isolated, institutionalized, and awkward. Neutral adjectives consist of educational, clinical, realistic, and helpful. The last portion asked students to rank how comfortable they would feel showing the respective photograph to a patient on a 5-point Likert scale. After viewing the traditional and natural photographs, students were shown the six different photographs together for comparison. They were then surveyed again on whether they would feel comfortable showing each photograph to a patient (yes or no) and invited to share their thoughts on the traditional and natural photographs within the survey. The final section of the survey queried additional demographic information to the known degree program. All questions were mandatory with the exception of the free response question. However, respondents could exit the survey at any time. This resulted in varying degree of completeness, from respondents that completed the whole survey to those that partially completed the questions in the first period of the study (prior to crossover). This resulted in differing sample sizes for different analyses.

### *Data Analysis*

Survey responses were coded and entered into a Microsoft Excel spreadsheet. Statistical analysis was then performed using STATA (v.13. College Station, TX). Statistical significance was assumed at a Type I error rate of 5%. The cross-over design was analyzed using multivariable mixed models: ordered logistic models for Likert scales and logistic models for the dichotomous adjective data. Analyses included evaluations of differences in perception between the traditional and natural photographs and whether these opinions differed between and within student groups and between students randomized to arm A and arm B.

This methodology allowed for the evaluation of both intra-individual effects and inter-individual effects. Intra-individual effects included the *treatment effect* (differences between traditional and natural photographs for the same person) and the *sequence effect* (differences due to the order the two types of

photographs were viewed). The inter-individual effect was the *period effect* (differences between overall first group of photographs viewed and second group of photographs viewed) (Senn *et al.* 2002).

## RESULTS

A total of 844 participants from the surveyed institutions were randomized, with 413 students randomized into arm A and 431 students into arm B. Of these, 549 participants (65%) completed the survey, including 267 students from arm A and 282 students from arm B (Figure 2). The survey was distributed to 2,578 medical students at UT Houston, Baylor College of Medicine, UTMB, and Virginia Commonwealth University, 537 genetic counseling students in North America, 787 dental students at UT School of Dentistry and Virginia Commonwealth University, and 1,167 nursing students at UT School of Nursing. Twelve percent of medical students, 40% of genetic counseling students, and 7% of dental and nursing students partially completed the survey. Ten percent of medical students, 36% of genetic counseling students, and 6% of dental and nursing students completed the entire survey.

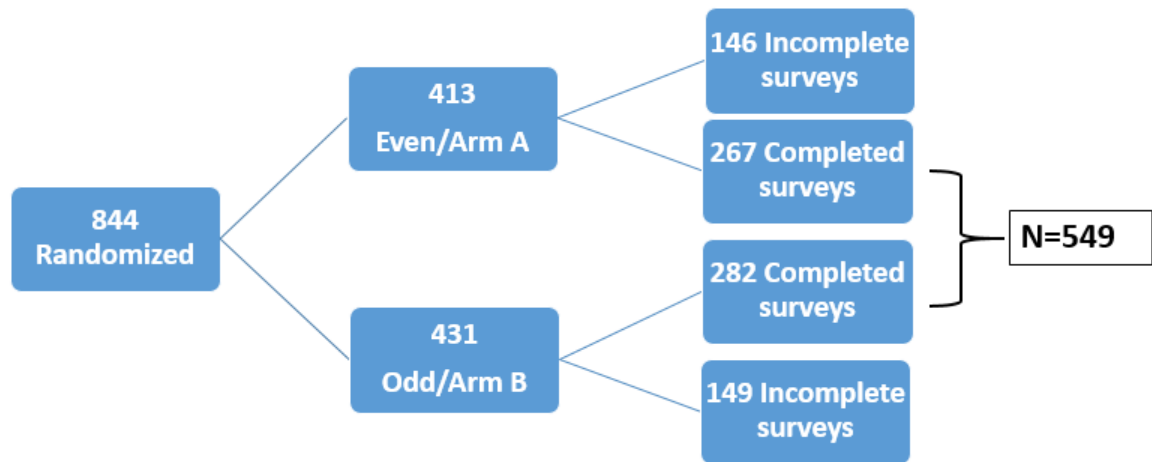


Figure 2: Participation Flowchart

844 students were randomized into arm A or arm B based on an even or odd day of birth. Out of the 413 students in arm A, 267 completed the survey. Out of the 431 students in arm b, 282 completed the survey.

### *Demographics*

The majority of respondents (>60%) were female and between 18-35 years of age. Forty-six percent of participants were medical students followed by genetic counseling (33.4%), nursing (12.2%), and dental students (8.3%). The two arms were similar with respect to gender, age, educational program, knowledge of an individual with one of the studied genetic conditions, and either a personal history or family member or close friend with a genetic condition with obvious physical manifestations (Table 1).

**Table 1. Demographics**

	Randomization Arm, n (%)	
	Arm A	Arm B
Gender (n=549)		
Female	190 (71)	210 (74)
Male	77 (29)	72 (25)
Age (n=549)		
18-25 years	180 (67)	184 (65)
26-30 years	68 (26)	76 (27)
31-35 years	14 (5)	15 (5)
36 years +	5 (2)	7 (3)
Program (n=649*)		
GC, n=217		
1st year	51 (50)	63 (55)
2nd year	51 (50)	51 (45)
Med, n=299		
1st year	34(25)	34 (21)
2nd year	36 (26)	43 (27)
3rd year	35 (26)	44 (27)
4th year	32 (23)	40 (25)
Nursing, n= 79		
1st year	28 (68)	19 (50)
2nd year	13 (32)	19 (50)
Dental, n=54		
1st year	13 (42)	9 (39)
2nd year	8 (26)	3 (13)
3rd year	6 (19)	4 (17)
4th year	4 (13)	7 (30)
Know individual with ...		
Marfan (n=652*)	56 (18)	44 (13)
CdL (n=612*)	5 (2)	9 (3)
Noonan (n=604*)	9 (3)	14 (5)
Personal genetic condition with obvious physical manifestations (n=549)	5 (2)	4 (1)
Family member or close friend with a genetic condition with obvious physical manifestations (n=549)	43 (16)	40 (14)

\*Sample sizes for each question differ as students completed the survey to varying degrees

*Treatment Effect-Significant differences between Traditional and Natural photographs*

Students indicated their perception of quality of life characteristics of individuals with Marfan, CdL, and Noonan syndrome after viewing the traditional and natural photographs using the QoLA. The most frequently reported QoLA scores were 2 and 3 for the traditional photographs, 3 and 4 for the natural Marfan syndrome photograph, and 2 and 3 for the natural CdL and Noonan syndrome photographs. Natural photographs were significantly more likely to be associated with higher Likert scores in comparison to the traditional photographs ( $p < 0.001$ ) for all Likert scale questions with the exception of the characteristics “attractiveness”, “competence”, and “intelligence” for the Noonan syndrome photographs (Table 2).

Students also reported whether the adjectives came to mind when viewing the traditional and natural photographs using the Perception Assessment (Table 2). Positive adjectives were more likely to be associated with the natural photographs than with the traditional photos ( $p < 0.001$ ). Conversely, the negative adjectives were more likely to be associated with the traditional rather than the natural photographs ( $p < 0.001$ ). The majority of students associated the neutral adjectives “educational” and “clinical” with the traditional images, although  $>50\%$  of students still reported that the term “educational” came to mind when viewing the natural photographs ( $p < 0.05$ ). “Helpful” and “realistic” were significantly more likely to be associated with the natural photos, although  $>50\%$  of students still reported that the term “realistic” came to mind when viewing the traditional images (Table 2).

**Table 2. Treatment Effect\*\*\*-Differences between Traditional and Natural Images**

	Marfan syndrome		Noonan syndrome		CdL syndrome	
	Traditional	Natural	Traditional	Natural	Traditional	Natural
<b>Quality of Life Assessment, median (IQR)</b>						
Quality of Life	3 (2 - 3)	4 (3 - 4)	2 (2 - 3)	3 (2 - 4)	2 (1 - 3)	3 (3 - 4)
Attractiveness <sup>†</sup>	2 (2 - 3)	3 (2 - 3)	2 (1 - 3)	2 (1 - 3)	2 (1 - 2)	2 (2 - 3)
Popularity	2 (2 - 3)	3 (3 - 4)	2 (1 - 3)	2 (2 - 3)	2 (1 - 3)	3 (2 - 3)
Competence <sup>†</sup>	3 (3 - 3)	3 (3 - 4)	2 (2 - 3)	2 (2 - 3)	2 (2 - 3)	3 (2 - 3)
Intelligence <sup>†</sup>	3 (3 - 4)	3 (3 - 4)	2 (2 - 3)	2 (2 - 3)	2 (2 - 3)	3 (2 - 3)
Hardworking	3 (3 - 4)	3 (3 - 4)	3 (2 - 3)	3 (2 - 3)	3 (2 - 3)	3 (3 - 4)
Communicativeness	3 (2 - 3)	3 (3 - 4)	2 (2 - 3)	3 (2 - 3)	2 (2 - 3)	3 (2 - 3)
<b>Perception Assessment, n (%)</b>						
<b>Positive Adjectives</b>						
Creative	65 (10.8)	304 (49.8)	30 (5.2)	241 (41.5)	36 (6.2)	177 (30.0)
Respectful	86 (14.3)	314 (51.4)	63 (11.0)	345 (59.4)	57 (9.9)	346 (58.6)
Beautiful	50 (8.3)	241 (39.4)	38 (6.6)	230 (39.6)	48 (8.3)	159 (27.0)
Individualistic	178 (29.6)	478 (78.2)	122 (21.3)	394 (67.8)	124 (21.5)	380 (64.4)
Flattering	7 (1.2)	170 (27.8)	10 (1.7)	169 (29.1)	10 (1.7)	159 (27.0)
Artistic	41 (6.8)	207 (33.8)	24 (4.2)	253 (43.6)	34 (5.9)	188 (31.9)
Relatable	72 (12.0)	392 (64.2)	55 (9.6)	274 (47.2)	41 (7.1)	302 (51.2)
<b>Negative Adjectives</b>						
Degrading	376 (62.5)	48 (7.9)	345 (60.1)	48 (8.3)	299 (51.7)	30 (5.1)
Humiliating	388 (54.5)	52 (8.5)	367 (63.9)	44 (7.6)	288 (49.8)	44 (7.5)
Neglected	217 (36.1)	44 (7.2)	199 (34.7)	48 (8.3)	244 (42.2)	50 (8.5)
Depressing	276 (45.9)	42 (6.9)	282 (49.1)	73 (12.6)	342 (59.2)	72 (12.2)
Isolated	349 (58.0)	74 (12.1)	347 (60.5)	100 (17.2)	347 (60.0)	153 (25.9)
Institutionalized	262 (43.5)	39 (6.4)	241 (42.0)	68 (11.7)	265 (45.9)	44 (7.5)
Awkward	466 (77.4)	231 (37.8)	399 (69.5)	142 (24.4)	323 (55.9)	134 (22.7)
<b>Neutral Adjectives</b>						
Educational*	383 (63.6)	308 (50.4)	363 (63.2)	338 (58.2)	362 (62.6)	335 (56.8)
Clinical	507 (84.2)	236 (38.6)	475 (82.8)	252 (43.4)	483 (83.6)	229 (38.8)
Realistic	307 (51.0)	469 (76.8)	293 (51.1)	440 (75.7)	308 (53.3)	450 (76.3)
Helpful	233 (38.7)	328 (53.7)	211 (36.8)	320 (55.1)	203 (35.1)	315 (53.4)

For the Quality of Life Assessment, median and interquartile ranges are listed for each characteristic. For the perception assessment, the number and percentage of students who selected that yes, the adjective came to mind when viewing each photograph are noted. \*\*\* = All treatment effects were significant at  $p < 0.001$  unless otherwise noted. \* = Treatment effect where  $p < 0.05$  for “educational” for Noonan and CdL syndrome. † = No significant treatment effect was seen for “attractiveness”, “competence”, and “intelligence” for Noonan syndrome.

*Sequence Effect- Significant Differences in Perception between Arm A and Arm B*

Students perceptions of quality of life characteristics differed between arm A and arm B ( $p < 0.001$ ). Generally, students in arm A, who initially viewed the traditional photos, had higher traditional and natural median QoLA scores and/or a more positively shifted interquartile range in comparison to students randomized to arm B (Table 3).

A higher percentage of students in arm A reported that the positive adjectives came to mind when viewing the traditional and natural photographs in comparison to the students in arm B ( $p < 0.05$ ). The only exception was “artistic” which had no significant difference between the two arms for any of the traditional or natural photographs. In contrast, generally, a lower percentage of students in arm A reported that negative adjectives came to mind when viewing the traditional and natural photographs in comparison to students in arm B ( $p < 0.05$ ). The sole exception was the adjective “neglected” which had no significant difference between the two arms for all of the photos evaluated by students.

A lower percentage of students in arm A in comparison to students in arm B thought that “clinical” came to mind when viewing the traditional and natural photographs, while a higher percentage of students in arm A found that “realistic” and “helpful” came to mind. No significant difference was seen between the two arms for the term “educational” (Table 3).

**Table 3. Sequence and Period Effect**

	Marfan syndrome					
Photograph	Traditional		Natural		Sequence effect	Period effect
Sequence	Trad --> Nat	Nat --> Trad	Trad --> Nat	Nat --> Trad		
<b>Quality of Life Assessment, median (IQR)</b>						
Quality of Life	3 (2 - 4)	3 (2 - 3)	4 (3 - 4)	3 (3 - 4)	***	NS
Attractiveness	2 (2 - 3)	2 (1 - 3)	3 (2 - 3)	3 (2 - 3)	***	NS
Popularity	3 (2 - 3)	2 (2 - 3)	3 (3 - 4)	3 (2 - 3)	***	NS
Competence	3 (3 - 4)	3 (2 - 3)	3 (3 - 4)	3 (3 - 4)	***	***
Intelligence	3 (3 - 4)	3 (2 - 3)	3 (3 - 4)	3 (3 - 4)	***	***
Hardworking	3 (3 - 4)	3 (2 - 3)	3 (3 - 4)	3 (3 - 4)	***	***
Communicativeness	3 (3 - 4)	3 (2 - 3)	4 (3 - 4)	3 (3 - 4)	***	***
<b>Perception Assessment, n (%)</b>						
<u>Positive Adjectives</u>						
Creative	39 (13)	26 (9)	160 (58)	144 (43)	**	NS
Respectful	59 (19)	27 (9)	181 (66)	133 (40)	***	NS
Beautiful	35 (11)	15 (6)	137 (50)	104 (31)	*	NS
Individualistic	121 (39)	57 (20)	213 (78)	265 (79)	**	NS
Flattering	3 (1)	4 (1)	116 (42)	54 (16)	**	NS
Artistic	22 (7)	19 (7)	105 (38)	102 (30)	NS	NS
Relatable	46 (15)	26 (9)	214 (78)	178 (53)	**	***
<u>Negative Adjectives</u>						
Degrading	169 (54)	207 (72)	11 (4)	37 (11)	***	NS
Humiliating	183 (59)	205 (71)	14 (5)	38 (11)	*	NS
Neglected	125 (40)	92 (32)	13 (8)	31 (9)	NS	**
Depressing	136 (44)	140 (48)	13 (8)	29 (9)	**	*
Isolated	179 (57)	170 (59)	17 (6)	57 (17)	***	***
Institutionalized	117 (37)	145 (50)	15 (6)	24 (7)	**	*
Awkward	254 (81)	212 (73)	42 (15)	189 (56)	***	***
<u>Neutral Adjectives</u>						
Educational	193 (62)	190 (66)	158 (58)	150 (45)	NS	NS
Clinical	254 (81)	253 (88)	80 (29)	156 (46)	***	NS
Realistic	174 (56)	133 (46)	234 (85)	235 (70)	**	NS
Helpful	131 (42)	102 (35)	185 (68)	143 (42)	**	**

**Table 3. Sequence and Period Effect (continued)**

	Noonan syndrome					
Photograph	Traditional		Natural		Sequence effect	Period effect
Sequence	Trad --> Nat	Nat --> Trad	Trad --> Nat	Nat --> Trad		
<b>Quality of Life Assessment, median (IQR)</b>						
Quality of Life	3 (2 - 3)	2 (2 - 3)	3 (3 - 4)	3 (2 - 3)	***	NS
Attractiveness	2 (2 - 3)	2 (1 - 2)	2 (2 - 3)	2 (1 - 2)	***	NS
Popularity	2 (2 - 3)	2 (1 - 2)	3 (2 - 3)	2 (2 - 3)	***	NS
Competence	3 (2 - 3)	2 (2 - 3)	3 (2 - 3)	2 (2 - 3)	***	NS
Intelligence	3 (2 - 3)	2 (2 - 3)	2 (2 - 3)	2 (2 - 3)	***	NS
Hardworking	3 (2 - 3)	2 (2 - 3)	3 (3 - 3)	3 (2 - 3)	***	***
Communicativeness	3 (2 - 3)	2 (2 - 3)	3 (2 - 3)	3 (2 - 3)	***	NS
<b>Perception Assessment, n (%)</b>						
<u>Positive Adjectives</u>						
Creative	20 (7)	10 (4)	132 (49)	109 (35)	**	NS
Respectful	44 (15)	19 (8)	189 (51)	156 (50)	***	NS
Beautiful	23 (8)	15 (5)	129 (48)	101 (32)	*	NS
Individualistic	79 (27)	43 (15)	198 (74)	196 (63)	***	NS
Flattering	7 (2)	3 (1)	112 (42)	57 (18)	**	NS
Artistic	17 (6)	7 (3)	121 (45)	132 (42)	NS	NS
Relatable	32 (11)	23 (8)	162 (60)	112 (36)	***	*
<u>Negative</u>						
Degrading	146 (50)	199 (70)	14 (5)	34 (11)	***	NS
Humiliating	167 (57)	200 (71)	16 (6)	28 (9)	**	NS
Neglected	98 (34)	101 (36)	16 (6)	32 (10)	NS	NS
Depressing	127 (44)	155 (55)	30 (11)	43 (14)	*	NS
Isolated	166 (57)	181 (64)	32 (12)	68 (22)	**	NS
Institutionalized	110 (38)	131 (46)	21 (8)	47 (15)	**	NS
Awkward	189 (65)	210 (74)	49 (18)	93 (30)	***	NS
<u>Neutral</u>						
Educational	189 (65)	174 (61)	165 (62)	173 (55)	NS	NS
Clinical	233 (80)	242 (86)	90 (34)	162 (52)	***	NS
Realistic	157 (54)	136 (48)	221 (83)	219 (70)	**	NS
Helpful	128 (44)	83 (29)	165 (62)	155 (50)	***	NS

**Table 3. Sequence and Period Effect (continued)**

	CdL syndrome					
Photograph	Traditional		Natural		Sequence effect	Period effect
Sequence	Trad --> Nat	Nat --> Trad	Trad --> Nat	Nat --> Trad		
<b>Quality of Life Assessment</b> , median (IQR)						
Quality of Life	2 (2 - 3)	2 (1 - 3)	3 (3 - 4)	3 (2 - 3)	***	**
Attractiveness	2 (1 - 2)	2 (1 - 2)	3 (2 - 3)	2 (2 - 3)	***	**
Popularity	2 (2 - 3)	2 (1 - 2)	3 (2 - 3)	2 (2 - 3)	***	**
Competence	2 (2 - 3)	2 (1 - 3)	3 (3 - 4)	3 (2 - 3)	***	NS
Intelligence	2 (2 - 3)	2 (1 - 3)	3 (3 - 3)	3 (2 - 3)	***	NS
Hardworking	2 (2 - 3)	2 (1 - 3)	3 (3 - 4)	3 (3 - 4)	***	***
Communicativeness	2 (2 - 3)	2 (1 - 3)	3 (3 - 4)	3 (2 - 3)	***	NS
<b>Perception Assessment</b> , n (%)						
<u>Positive Adjectives</u>						
Creative	21 (7)	15 (5)	106 (39)	71 (22)	**	NS
Respectful	35 (12)	22 (8)	187 (69)	159 (53)	***	NS
Beautiful	28 (10)	20 (7)	86 (32)	73 (23)	*	NS
Individualistic	79 (27)	45 (16)	187 (69)	193 (61)	**	NS
Flattering	7 (2)	3 (1)	104 (38)	55 (17)	**	NS
Artistic	20 (7)	14 (5)	99 (37)	89 (28)	NS	NS
Relatable	20 (7)	21 (7)	178 (66)	124 (39)	**	***
<u>Negative Adjectives</u>						
Degrading	136 (46)	163 (57)	6 (2)	24 (8)	***	NS
Humiliating	139 (47)	149 (52)	14 (5)	30 (9)	*	NS
Neglected	129 (44)	115 (40)	14 (5)	36 (11)	NS	**
Depressing	168 (57)	174 (61)	21 (8)	51 (16)	**	*
Isolated	176 (60)	171 (60)	32 (12)	121 (38)	***	***
Institutionalized	128 (44)	137 (48)	11 (4)	33 (10)	**	*
Awkward	159 (54)	164 (58)	34 (13)	100 (31)	***	***
<u>Neutral Adjectives</u>						
Educational	184 (63)	178 (63)	162 (60)	173 (54)	NS	NS
Clinical	236 (81)	247 (87)	82 (30)	147 (46)	***	NS
Realistic	164 (56)	144 (51)	224 (83)	226 (71)	**	NS
Helpful	109 (37)	94 (33)	171 (46)	144 (45)	**	**

\* = p&lt;0.05

\*\* = p&lt;0.01

\*\*\* = p&lt;0.001

NS = not significant

### *Period Effect*

There were some significant differences between student responses in period 1 compared to the responses in period 2. Examining the QoLA responses revealed a significant difference between the two periods for “competence”, “intelligence”, “hardworking”, and “communicativeness” for the Marfan photographs, “hardworking” for the Noonan photographs, and “attractiveness”, “popularity”, “hardworking”, and “quality of life” for the CDL photographs ( $p < 0.01$ , Table 3).

For the positive adjectives within the PA, the only significant difference between the two periods was for the term “relatable” ( $p < 0.05$ ). This difference was seen across all images. Greater variability was observed when evaluating for a period effect in the negative adjectives. For the Marfan photographs, significant differences between both periods for the terms neglected, depressing, isolated, institutionalized, and awkward were identified ( $p < 0.05$ ). For the CdL photographs, there were significant differences between period 1 and period 2 for the terms “neglected”, “depressing”, “isolated”, “institutionalized”, and “awkward” ( $p < 0.05$ ). Evaluating the neutral adjectives revealed significant differences between both periods for the term “helpful” for only the Marfan and CdL photographs ( $p < 0.01$ ). No significant differences between period 1 and period 2 were observed for the negative or neutral adjectives for the Noonan photographs. These results are summarized in Table 3.

### *Differences between Student Groups*

The responses of medical, nursing, and dental students were compared to the responses of genetic counseling students to identify differences in perception between participants from different healthcare programs. For the most part, there were no differences between the genetic counseling students and the other three students groups with respect to the QoLA or the PA items. However, a few significant differences did show up that varied by the student group and the condition being viewed (Table 4). When the difference was statistically significant ( $p < 0.05$ ), the medical, nursing and dental students assigned lower QoL scores compared to the genetic counseling students given that they were viewing the same photographs in the same period. For the PA responses, when differences were

identified, nursing students were significantly more likely to select that the positive and negative adjectives came to mind when viewing the images in comparison to genetic counseling students while medical and dental students were less likely to select these adjectives ( $p < 0.05$ ).

**Table 4. Odds Ratio for QoLA and PA Between Genetic Counseling, Medical, Nursing, and Dental Students**

	Marfan syndrome					
	Medical	p-value	Nursing	p-value	Dental	p-value
<b>Quality of Life Assessment</b>						
Quality of Life	0.63	**	0.51	**	0.32	***
Attractiveness	0.91	NS	0.73	NS	0.46	*
Popularity	0.93	NS	0.63	NS	0.61	NS
Competence	0.98	NS	1.13	NS	0.44	*
Intelligence	1.06	NS	1.49	NS	0.61	NS
Hardworking	1.36	NS	2.43	**	0.90	NS
Communicativeness	0.89	NS	1.16	NS	0.53	*
<b>Perception Assessment</b>						
<b>Positive Adjectives</b>						
Creative	0.89	NS	2.20	*	1.77	NS
Respectful	1.21	NS	3.26	***	1.74	NS
Beautiful	0.82	NS	3.38	**	2.58	NS
Individualistic	1.23	NS	4.18	***	2.93	**
Flattering	0.47	*	0.75	NS	0.36	*
Artistic	0.90	NS	2.04	NS	0.75	NS
Relatable	0.81	NS	0.95	NS	0.66	NS
<b>Negative Adjectives</b>						
Degrading	0.58	**	0.81	NS	0.52	*
Humiliating	0.57	**	1.29	NS	0.78	*
Neglected	0.98	NS	3.36	**	1.92	NS
Depressing	0.70	NS	2.47	**	0.95	NS
Isolated	0.66	*	1.53	NS	1.19	NS
Institutionalized	0.56	*	1.39	NS	0.67	NS
Awkward	0.99	NS	1.87	*	1.30	NS
<b>Neutral Adjectives</b>						
Educational	0.59	*	0.92	NS	0.43	*
Clinical	0.81	NS	1.20	NS	0.86	NS
Realistic	0.64	NS	0.88	NS	0.61	NS
Helpful	0.71	NS	1.53	NS	1.02	NS

**Table 4. Odds Ratio for QoLA and PA Between Genetic Counseling, Medical, Nursing, and Dental Students (continued)**

	Noonan syndrome					
	Medical	p-value	Nursing	p-value	Dental	p-value
<b>Quality of Life Assessment</b>						
Quality of Life	0.54	***	0.59	*	0.32	***
Attractiveness	0.60	**	0.93	NS	0.40	**
Popularity	0.61	*	0.62	NS	0.50	*
Competence	0.61	*	1.54	NS	0.39	**
Intelligence	0.73	NS	2.19	*	0.44	*
Hardworking	1.06	NS	2.10	*	0.44	*
Communicativeness	0.66	*	1.43	NS	0.38	**
<b>Perception Assessment</b>						
<b>Positive Adjectives</b>						
Creative	0.62	NS	2.95	**	1.15	NS
Respectful	0.70	NS	1.29	NS	0.84	NS
Beautiful	0.50	NS	3.01	*	1.85	NS
Individualistic	0.68	NS	5.00	***	1.68	NS
Flattering	0.45	**	0.95	NS	0.50	NS
Artistic	0.69	NS	2.42	*	1.42	NS
Relatable	0.67	NS	1.01	NS	1.15	NS
<b>Negative Adjectives</b>						
Degrading	0.51	**	1.03	NS	0.43	*
Humiliating	0.61	*	1.11	NS	0.69	NS
Neglected	1.04	NS	3.33	***	2.87	**
Depressing	0.91	NS	2.13	*	1.09	NS
Isolated	1.1	NS	2.09	*	1.34	NS
Institutionalized	0.81	NS	1.43	NS	0.94	NS
Awkward	1.02	NS	1.91	NS	1.52	NS
<b>Neutral Adjectives</b>						
Educational	0.55	*	1.09	NS	0.38	*
Clinical	0.84	NS	0.97	NS	0.61	NS
Realistic	0.61	NS	1.19	NS	0.31	*
Helpful	0.54	*	2.17	NS	0.32	*

**Table 4. Odds Ratio for QoLA and PA Between Genetic Counseling, Medical, Nursing, and Dental Students (continued)**

	CdL syndrome					
	Medical	p-value	Nursing	p-value	Dental	p-value
<b>Quality of Life Assessment</b>						
Quality of Life	0.70	*	0.85	NS	0.41	**
Attractiveness	0.77	NS	0.82	NS	0.34	**
Popularity	1.07	NS	0.83	NS	0.43	*
Competence	0.99	NS	1.83	*	0.35	**
Intelligence	0.93	NS	2.80	**	0.70	*
Hardworking	1.27	NS	1.89	*	0.66	NS
Communicativeness	0.92	NS	2.17	**	0.39	**
<b>Perception Assessment</b>						
<b>Positive Adjectives</b>						
Creative	1.15	NS	4.65	**	1.49	NS
Respectful	0.97	NS	2.19	*	1.34	NS
Beautiful	1.05	NS	4.27	**	3.45	*
Individualistic	0.94	NS	5.19	***	1.27	NS
Flattering	0.42	**	0.58	NS	0.32	*
Artistic	1.01	NS	2.23	*	0.99	NS
Relatable	0.46	*	0.63	NS	0.58	NS
<b>Negative Adjectives</b>						
Degrading	0.55	**	1.63	NS	0.77	NS
Humiliating	0.69	NS	1.77	NS	0.98	NS
Neglected	1.04	NS	2.57	**	2.52	*
Depressing	0.80	NS	3.25	**	1.72	NS
Isolated	0.89	NS	1.78	NS	2.45	*
Institutionalized	0.68	NS	1.90	NS	0.77	NS
Awkward	0.86	NS	1.92	*	1.47	NS
<b>Neutral Adjectives</b>						
Educational	0.64	*	0.99	NS	0.65	NS
Clinical	1.05	NS	1.33	NS	0.83	NS
Realistic	0.64	NS	1.01	NS	0.37	*
Helpful	0.77	NS	1.37	NS	0.54	NS

\* = p&lt;0.05

\*\* = p&lt;0.01

\*\*\* = p&lt;0.001

NS = not significant

*Differences within Student Groups*

As this study did not have a large sample of nursing and dental students, differences in perception within these degree programs could not be examined. The perceptions of first year and second year genetic counseling students were similar.

The responses of second through fourth year medical students were compared to first year medical students to identify potential differences in perception within a degree program. The majority of responses were similar between medical students. Significant differences were seen between medical students' Marfan QoLA responses. Second year students reported higher QoLA scores for "competence", "intelligence", and "communicativeness" while third year medical students had lower QoLA scores for "intelligence" in comparison to first year students ( $p < 0.05$ ). Second year medical students had lower QoLA scores for "attractiveness" for the Noonan photographs in comparison to first, second, and third year students. For the CdL photographs, third and fourth year students had lower QoLA scores for "attractiveness" than first through second year students ( $p < 0.05$ ). Third year medical students, had lower QoLA scores for "popularity" and "hardworking" than their counterparts. PA differences were seen for "respectful" for Noonan and for "awkward" and "educational" for Noonan and CdL ( $p < 0.05$ , Table 5).

**Table 5. Odds Ratios for QoLA and PA for 2nd through 4th year Medical Students compared to 1st year Medical Students**

	Marfan syndrome			Noonan syndrome			CdL syndrome		
	2nd	3rd	4th	2nd	3rd	4th	2nd	3rd	4th
<b>Quality of Life Assessment</b>									
Quality of Life	1.21	0.88	1.27	1.12	0.56	1.01	0.95	0.56	0.53
Attractiveness	0.98	0.50	0.71	0.64	0.45	<b>0.43</b>	0.86	<b>0.42</b>	<b>0.39</b>
Popularity	1.20	0.54	1.07	0.62	0.56	0.80	0.56	<b>0.34</b>	0.54
Competence	<b>2.32</b>	0.57	1.04	1.10	0.52	0.77	1.14	0.64	0.48
Intelligence*	<b>2.21</b>	<b>0.42</b>	0.98	1.16	0.47	0.74	1.47	0.51	0.47
Hardworking*	<b>2.44</b>	0.62	1.32	1.94	0.58	0.73	1.78	<b>0.45</b>	0.55
Communicativeness*	<b>1.95</b>	0.58	1.05	1.25	0.61	0.71	1.17	0.53	0.62
<b>Perception Assessment</b>									
<b>Positive Adjectives</b>									
Creative	0.86	0.82	0.47	1.12	0.91	0.98	1.40	1.61	1.44
Respectful*	1.24	0.96	1.06	<b>3.47</b>	1.46	1.55	2.60	1.69	1.28
Beautiful	1.21	0.39	0.78	1.98	0.27	0.81	1.64	0.70	1.15
Individualistic	1.45	0.96	0.75	2.52	1.11	1.07	1.79	1.37	1.47
Flattering	1.13	1.15	0.90	2.31	1.25	1.79	2.61	2.13	2.18
Artistic	1.29	0.73	0.70	1.14	0.66	0.51	2.04	0.87	1.55
Relatable	1.61	1.10	1.22	1.34	0.68	1.25	1.26	0.63	1.30
<b>Negative Adjectives</b>									
Degrading	1.12	1.69	1.63	1.30	1.17	0.83	0.88	1.20	1.62
Humiliating	0.84	1.06	1.34	2.16	2.10	1.92	1.03	1.58	1.66
Neglected	1.47	1.57	1.41	0.96	1.32	1.16	1.96	2.11	1.78
Depressing	1.35	1.85	1.82	1.86	2.19	1.55	1.30	1.59	2.09
Isolated	0.83	0.95	1.28	1.78	1.15	1.84	1.09	1.14	1.49
Institutionalized	1.38	1.22	1.33	1.63	1.85	1.84	1.37	1.36	1.31
Awkward	1.91	1.66	1.53	<b>5.49</b>	<b>3.41</b>	<b>3.38</b>	<b>2.85</b>	1.84	<b>2.77</b>
<b>Neutral Adjectives</b>									
Educational	2.05	1.28	1.55	<b>3.99</b>	2.36	2.05	<b>3.36</b>	1.74	1.99
Clinical	1.40	0.85	1.24	1.64	0.92	1.34	1.30	0.56	1.59
Realistic	1.50	0.88	0.74	2.81	0.75	0.81	1.35	0.61	0.72
Helpful	2.23	2.11	1.29	4.31	1.84	3.53	1.90	1.36	1.28

Bold odds ratio values indicate significant difference between medical students within that year and first year medical students,  $p < 0.05$

### *Free Response*

At the end of the survey, 227 students elected to provide open ended comments on the traditional and natural photographs. Several themes that emerged were: 1) natural photographs are educational and beneficial for healthcare students, 2) reinforcement of the adjectives identified within the PA/negative associations with traditional images, 3) change in students' perception of traditional photographs after exposure to natural photos, and 4) utility of natural photographs for patients with these genetic disorders and their families.

The following quotations demonstrate the above themes:

Natural photographs are educational and beneficial for healthcare students:

- “The natural photographs are just as educational as the traditional and don't elicit a negative or uncomfortable feeling about the person in the photograph”
- “There's a non-exploitative way to clinically present individuals affected with genetic conditions, and this conversation is very much needed to challenge the antiquated medical model of genetics/dysmorphology education.”

Reinforcement of the adjectives portion of the questionnaire/negative associations with traditional images:

- “Traditional clinical pictures, such as those frequently seen in clinical textbooks, are degrading and immediately depict a negative sense of the condition”
- “The tradition[al] (*sic*) photographs present the patients as dehumanized clinical subjects, as if these individuals are only present for scrutiny and learning purposes. The natural photos show the patients engaged in normal, everyday life activities, which I think helps to humanize both the subject and the genetic condition. When I see the first set of photos (the traditional photos), I don't see a person but rather a clinical presentation of a disorder; I see stigma and shame. When I look at the natural photos of the patients, I see unique individuals living full, unconfined lives; in these photos I find joy, hope, and individualism.”

Change in students' perceptions of traditional photographs after exposure to natural photographs:

- “The traditional photos seemed okay, but once I saw the natural photos the traditional ones seem significantly more awkward.”

Utility of natural photographs for patients with these genetic disorders and their families:

- “Natural photographs can give patients a better idea of what a child with this disorder will actually be like. The traditional clinical pictures can be almost disturbing and degrading to individuals with the disorder.”

## DISCUSSION

Photographs are integral to many facets of our lives. Within an educational setting, images are crucial aspects of healthcare students' training to allow them to learn certain dysmorphic features and characteristic presentations of specific genetic conditions. This study demonstrates that the pictures of individuals with visible genetic conditions used can affect students' perceptions of the depicted individual's quality of life. As stated by Dr. Nicolas Franchitto et al (2008), "To take a photograph of a person is to lay bare their identity to the eyes of others. The photograph generates an ambiguous relationship with the idea of identity. It can in turn lay it bare, exploit it, reveal and embody it. It creates an image which takes on its own existence separately from the person portrayed." While the purpose of medical photographs is to emphasize subjects' features, they have the ability to impart much more to viewers.

The results of this study provide evidence that the type of photograph presented to students significantly impacts their perception of the subject's quality of life. Natural photographs were significantly more likely to be associated with higher Likert scores in comparison to the traditional photographs. These differences were significant ( $p < 0.001$ ) for all QoLA characteristics with the exception of attractiveness, competence, and intelligence for the Noonan syndrome photographs. This could be due to the fact that the natural Noonan photograph was a close up portrait of the subject's face emphasizing the individual's facial features. Conversely, the traditional Noonan photograph showed the subject's entire body highlighting the individual's nudity perhaps detracting from the subject's dysmorphic features. Individuals with Noonan syndrome can have noticeable to subtle facial features due to variable expressivity (Allanson et al, 2016). The more striking facial features in the natural Noonan photograph could have impacted students' perceptions and may contribute to the lack of significance found for those three terms.

The PA revealed that positive adjectives were more commonly associated with the natural photographs, while negative adjectives were more commonly associated with the traditional photographs. These responses further emphasize the differences in perception students draw from these

two types of images. While more students (63%) reported that the term “educational” came to mind when viewing the traditional compared to the natural photographs, the latter images were still considered “educational” by slightly more than half of respondents. Wood *et al.* (2013) demonstrated that there was no difference in students learning and that they are able to remember important concepts associated with genetic conditions regardless of whether a traditional or natural photograph accompanied a textbook-like entry. Students surveyed within this study seemed to agree with that finding. One student stated, “I love the natural photographs because they are still educational. They enable clinicians to learn about the physical features of these conditions while simultaneously being able to appreciate the positive aspects of these diagnoses. The natural photographs show these kids as individuals, not just as the subject of a clinical photograph.” Many other participants found that the natural photos were equally as educational as the traditional images without imparting a negative connotation about the photographic subjects’ quality of life. Interestingly, the majority of students found the natural images to be “realistic” and “helpful” while the term “clinical” was associated with the traditional photographs. The word clinical can have a negative connotation and was likely applied to the traditional photographs because the majority of healthcare students may have thought of similar images commonly observed within textbooks as part of their training. The wide use of traditional photographs within students’ training allows for the normalization of this type of photograph and the stark portrayal of the photographic subject.

While the same overall trends were present in students who were randomized to arm A and arm B, there were differences between the responses of students in each arm. Natural photos were still associated with higher quality of life perceptions and more positive adjectives, but these scores were generally shifted higher or more positively for students in arm A who saw the traditional images before the natural ones. This sequence effect provides evidence for a carryover effect, where the type of photograph students initially observed impacted their scoring for questions on the second set of photographs. Most studies which employ a cross-over design try to minimize the carryover effect as much as possible. However, for our study we were interested in identifying whether the initial

photograph anchors perception in the students, giving rise to a carryover effect. As such, this carryover effect was not perceived as a disruptive bias but rather as an outcome of interest within our study.

Anchoring is a cognitive bias where an individual's beliefs and judgements of values are influenced by anchors, starting points set by a previously considered standard. This effect was observed by providing students with specific anchors (traditional or natural photographs in the first period) and asking them to compare this anchor value to a different type of image (Tversky and Kahneman, 1974). Students in arm A were anchored by the traditional photographs. Students randomized to arm B were anchored by the natural photographs. Therefore, when observing the natural photographs, students in arm A provided higher QoLA scores and more strongly associated the images with positive adjectives, since the images were perceived as relatively more positive than the anchored traditional photograph. Conversely, students in arm B gave neutral scores to the natural images and then preceded to give relatively lower scores to the subsequently viewed traditional photos. This reasoning was reflected in the response section, as several students commented that initially they were unsure what to make of the natural photographs until they were able to compare them to the traditional photographs. While overall, the traditional photographs continued to be associated with negative perceptions and the natural photographs with positive perceptions, the magnitude of their perception was dictated by the type of photograph students initially viewed.

Differences in perception were also observed between each type of student groups. Genetic counseling students consistently rated the photographic subjects more positively, while medical, nursing, and dental students tended to have more negative perceptions. These trends could be due to the differences in educational focus between the four degree programs. Each healthcare students group's training includes a different emphasis on what is important, particularly within their genetics training. Genetic counseling students are trained to make both a genetic risk assessment, as well as a psychosocial risk assessment, while for medical students, the priority is making accurate diagnoses. These differences in perception could also be due to the fact that certain personalities are drawn to different healthcare

programs and career paths. However it should be noted that the overall trends observed for both treatment and sequence effects were present across student groups.

### *Educational Implications*

Our research demonstrates that student's quality of life perceptions of individuals with visible genetic conditions is impacted by the type of photograph presented and that the traditional photographs commonly found within textbooks are associated with negative perceptions in comparison to natural photographs. This finding has the capability to transform healthcare education by changing the images used to teach students characteristic presentations of genetic conditions. Based on the study by Nicole Wood (2013), natural photographs are just as educational as traditional photographs. However, they have the added benefit of imparting a sense of that individual that is sorely lacking in the images currently used in clinical education. Studies have demonstrated how implicit biases contribute to healthcare disparities and can impact patient care. (Blair *et al.* 2011). The images used for student instruction could contribute to these unconscious biases. Replacing traditional photographs with natural images could potentially neutralize negative biases, diminish stigma, and increase awareness among future healthcare providers of the perceptions they draw, often subconsciously, from textbook images.

In order for this transition to take place though, standardization of natural photos is necessary. This process can be difficult as it is important to ensure that these photographs clearly depict the clinical features of individuals with genetic conditions while being aware of how the subject is presented. It is also vital that each condition is portrayed accurately, mindful not to impart or suggest a more positive prognosis than typical such that a skewed view of the condition is taken. Photographs can be selected that highlight the subject's individuality and humanity in addition to their clinical features. Additionally, a photograph of a nude subject is not necessary to demonstrate every clinical feature. Select features can be demonstrated through photographs without requiring the nudity of the photographic subject.

### *Study Limitations*

One limitation of this study was the low response rate from each student group with the exception of genetic counseling students. Only a small portion of students who were given the opportunity to participate in the study responded to the questionnaire. While the sample size may affect the generalizability of the study results, crossover studies require lower sample sizes than other designs to achieve the same power and error risk (Wellek *et al*, 2012). Despite the benefits of this design, the small sample size largely prevented the investigation of differences in perception within a degree program to assess differences by students' academic year. Additionally this survey has not been validated.

This study did not use a washout period, or time between treatments, to prevent the introduction of anchoring biases into the proceeding periods. It is unclear how long of a time period would be necessary to allow the effects of the first type of photograph to dissipate. Moreover, in this instance, it was important to study the carryover effect and its impact on students' perceptions. In clinic, clinicians' experiences and perceptions of their patients result in a real time carryover and anchoring effect. However, it would be interesting for future research to see if the perceptions and anchoring effects persist over longer periods of time.

A period effect was also observed for some characteristics within this study. Differences between the first and second groups of photos observed could arise from the fact that students only saw a factual excerpt describing the three visible genetic conditions included within the study before viewing the first photograph within the questionnaire. The availability of this information before the introduction of the second type of photograph may have impacted students' responses. Furthermore, the differences between photographic subjects' ages and emphasis on their features may have also contributed to the observed period effect. For example, the traditional CdL image featured an infant, while the natural photograph featured an older child. Attempting to match the two types of photographs for characteristics that can be accounted for like age, gender, ethnicity, and severity of condition in future studies may remove the possibility of these biases.

*Research Recommendations*

Further research with medical, dental and nursing students to obtain a larger sample size is indicated to expand upon the findings of this study. Greater numbers would not only allow for the examination of differences between these healthcare programs but also for differences within each degree program and add to the data from this study. Continued qualitative research and detailed analysis of student's free response comments may help elucidate further avenues for study. Additionally, the analysis of student responses from different institutions with attention to the timing of the introduction of genetics material and of students' clinical rotations would be of interest. Investigation into other healthcare programs like physician's assistant, occupational and physical therapy, and dental hygiene programs can allow for broader generalizability. Furthermore, research into the perceptions of undergraduate students and healthcare professionals like physicians, practicing genetic counselors, dentists, and nurses can allow us to see where potential biases are introduced and if clinical experience impacts and stratifies providers' opinions.

## APPENDIX A: STUDY INVITATION

Dear healthcare student,

My name is Christina Falugi, and I'm a genetic counseling student at the University of Texas Health Science Center at Houston. You are invited to participate in my master's thesis, an educational project examining healthcare students' perceptions of individuals with visible genetic conditions from photographs.

This study will consist of a short online survey that should take no more than 10-15 min to complete. Participation in this survey is voluntary. Should you choose to participate, you will be asked some basic demographic questions and to respond to questions relating to three visible genetic conditions.

You are being asked to participate in this study because you are a student in the healthcare field. Choosing to participate in this study will enable us to better understand student learning concerning individuals with visible genetic conditions.

To protect your identity, no identifiable information will be collected. You can also choose to submit your e-mail address to be entered into a drawing for **one of three \$25 Visa gift cards** at the end of the survey. You will be asked for your email address upon completion of the survey.

If you are willing to participate, please follow the link:

<https://redcap.uth.tmc.edu/surveys/?s=hZforTr7XN>

If you have any further questions please feel free to contact me at [christina.falugi@uth.tmc.edu](mailto:christina.falugi@uth.tmc.edu).

Thank you for your time.

Sincerely,

Christina, Falugi, BS  
Genetic Counseling Intern II  
UT Health  
[christina.falugi@uth.tmc.edu](mailto:christina.falugi@uth.tmc.edu)

Sarah Elsea, Ph.D, FACMG  
Associate Professor  
Baylor College of Medicine  
Department of Molecular & Human Genetics  
[sarah.elsea@bcm.edu](mailto:sarah.elsea@bcm.edu)

## APPENDIX B: STUDENT QUESTIONNAIRE

1. What degree program are you currently enrolled in?

- ☐ Genetic Counseling
  - ☐ 1<sup>st</sup> year
  - ☐ 2<sup>nd</sup> year
- ☐ Nursing
  - ☐ Please specify your current degree program. Ex. Masters in Nursing, Doctorate, etc
  - ☐ What is your current year? Ex. 1<sup>st</sup> year, 2<sup>nd</sup> year, etc
- ☐ Dentistry
  - ☐ 1<sup>st</sup> year
  - ☐ 2<sup>nd</sup> year
  - ☐ 3<sup>rd</sup> year
  - ☐ 4<sup>th</sup> year
- ☐ Dental Hygiene
  - ☐ 1<sup>st</sup> year
  - ☐ 2<sup>nd</sup> year
- ☐ Medical School
  - ☐ 1<sup>st</sup> year
  - ☐ 2<sup>nd</sup> year
  - ☐ 3<sup>rd</sup> year
  - ☐ 4<sup>th</sup> year
- ☐ Other
  - ☐ Please specify your current degree program. Ex. Physician assistant school, etc
  - ☐ What is your current year? Ex. 1<sup>st</sup> year, 2<sup>nd</sup> year, etc
- ☐ Not currently enrolled [END SURVEY]

2. What institution are you currently enrolled in?

- ☐ UT Health Science Center
- ☐ UT Medical Branch
- ☐ Baylor College of Medicine
- ☐ Virginia Commonwealth University
- ☐ Other
  - ☐ Please specify your institution.

3. Is your day of birth on an even or odd day? For example, October 15<sup>th</sup> would be an odd day. This question is for randomization purposes.

- ☐ Even
- ☐ Odd

Please read the following descriptions of Marfan syndrome, Cornelia de Lange syndrome, and Noonan syndrome to provide you some background when assessing your perception of individuals with these disorders. Because you may not be familiar with these conditions, this information is solely to provide you with some context. You will not be tested on the information about these disorders.

### Marfan syndrome

Marfan syndrome is a connective tissue disorder affecting the skeletal, ocular, and cardiovascular systems. This disorder is caused by mutations in the Fibrillin 1 (*FBN1*) gene and is associated with a high degree of clinical variability. Individuals with Marfan syndrome are typically tall and slender and have arachnodactyly (elongated fingers and toes). Additional findings include pectus carinatum or excavatum, scoliosis, ectopia lentis (lens dislocation), aortic root dilation, mitral valve prolapse, and spontaneous pneumothorax. IQ is generally not affected although learning disability and/or hyperactivity can be seen.

### Cornelia de Lange syndrome

Cornelia de Lange syndrome (CdLS) is characterized by small stature, hirsutism (excessive hairiness), and characteristic facial features including synophrys (unibrow), arched eyebrows, long eyelashes, small widely spaced teeth, small upturned nose, and a high arched palate. Almost all individuals with CdLS have limb abnormalities ranging from complete absence of the forearms to micromelia (small hands). Individuals with CdLS often have severe to profound intellectual disability and IQs ranging from <30-102. CdLS is primarily caused by mutations in the *NIPBL* gene and rarely in the *SMC1A* and *SMC3* genes.

### Noonan syndrome

Noonan syndrome (NS) is characterized by short stature, congenital heart defects, and mild to moderate developmental delay. Physical characteristics can include low-set, posteriorly rotated ears, vivid blue or blue-green irises, widely-spaced eyes with epicanthal folds, a broad or webbed neck, low posterior hairline, superior pectus carinatum and inferior pectus excavatum (protruding or sunken chest), and cryptorchidism (undescended testicles) in males. This condition can be caused by mutations in a number of different genes with ~50% due to mutations in the *PTPN11* gene.

## Marfan syndrome

*Marfan syndrome image not included as permission was not granted.*

4. Do you know an individual with Marfan syndrome?

☐ Yes

☐ No

5. How would you rate an individual with Marfan syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5
Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

6. Does the above photograph bring the following adjectives to mind?

Creative	Yes	No
Degrading	Yes	No
Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No
Relatable	Yes	No
Clinical	Yes	No
Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

7. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable  
comfortable

Extremely

1

2

3

4

5

### Cornelia de Lange syndrome

*CdL syndrome image not included as permission was not granted by Elsevier.*

8. Do you know an individual with Cornelia de Lange syndrome?

☐ Yes

☐ No

9. How would you rate an individual with Cornelia de Lange syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5
Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

10. Which of the following adjectives does the above photograph bring to mind?

Creative	Yes	No
Degrading	Yes	No
Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No
Relatable	Yes	No
Clinical	Yes	No

Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

11. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable					Extremely comfortable
1	2	3	4	5	

### Noonan syndrome

*Noonan syndrome image not included as permission was not granted by Elsevier.*

12. Do you know an individual with Noonan syndrome?

- ☐ Yes  
☐ No

13. How would you rate an individual with Noonan syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5
Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

14. Which of the following adjectives does the above photograph bring to mind?

Creative	Yes	No
Degrading	Yes	No
Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No

Relatable	Yes	No
Clinical	Yes	No
Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

15. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable  
comfortable

Extremely

1 2 3 4 5

### Marfan syndrome



16. How would you rate an individual with Marfan syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5
Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

17. Which of the following adjectives does the above photograph bring to mind?

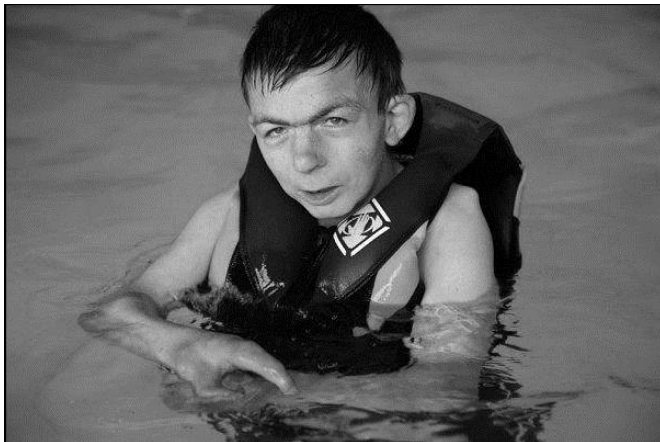
Creative	Yes	No
Degrading	Yes	No

Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No
Relatable	Yes	No
Clinical	Yes	No
Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

18. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable					Extremely
comfortable					
	1	2	3	4	5

#### Cornelia de Lange syndrome



19. How would you rate an individual with Cornelia de Lange syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5
Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

20. Which of the following adjectives does the above photograph bring to mind?

Creative	Yes	No
Degrading	Yes	No
Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No
Relatable	Yes	No
Clinical	Yes	No
Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

21. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable					Extremely
comfortable					
	1	2	3	4	5

#### Noonan syndrome



22. How would you rate an individual with Noonan syndrome after viewing the above photograph in terms of the following characteristics on a scale of 1 to 5? For example for attractiveness, 1=not at all attractive and 5=exceptionally attractive.

	Least				Most
Attractiveness	1	2	3	4	5
Popularity	1	2	3	4	5
Competence	1	2	3	4	5
Intelligence	1	2	3	4	5

Hardworking	1	2	3	4	5
Communicativeness	1	2	3	4	5
Quality of life	1	2	3	4	5

23. Which of the following adjectives does the above photograph bring to mind?

Creative	Yes	No
Degrading	Yes	No
Humiliating	Yes	No
Respectful	Yes	No
Beautiful	Yes	No
Educational	Yes	No
Depressing	Yes	No
Isolated	Yes	No
Flattering	Yes	No
Relatable	Yes	No
Clinical	Yes	No
Realistic	Yes	No
Institutionalized	Yes	No
Awkward	Yes	No

24. How comfortable would you feel showing this photograph to a patient on a scale of 1 to 5?

Not at all comfortable					Extremely
comfortable	1	2	3	4	5

## Traditional Photographs

*Marfan syndrome image not included as permission was not granted.*

*CdL and Noonan syndrome images not included as permission was not granted by Elsevier publisher.*

1.

2.

3.

## Natural Photographs



4.

5.

6.

25. Do you feel comfortable showing each of these photographs to a patient?

#1	Yes	No
#2	Yes	No
#3	Yes	No
#4	Yes	No
#5	Yes	No
#6	Yes	No

26. Please feel free to share your thoughts about the traditional photographs in comparison to the natural photographs.

27. What is your gender?

- ☐ Male
- ☐ Female

28. What is your age?

- ☐ 18-25 years
- ☐ 26-30 years
- ☐ 31-35 years
- ☐ 36 years and older

29. Do you have a genetic condition with obvious physical manifestations?

- ☐ Yes
- ☐ No

30. Do you have an immediate family member or a close friend with a genetic condition with obvious physical manifestations?

- ☐ Yes
- ☐ No

31. What is your ethnicity? Select all that apply.

- ☐ Caucasian (European, non-Hispanic, Middle East)
- ☐ African American
- ☐ Latino/Hispanic
- ☐ South East or North East Asian
- ☐ Pacific Islander
- ☐ Native American
- ☐ Other (Please specify) \_\_\_\_\_.

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**VITA**

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