

---

# Psychological Considerations: Visible Distinctions and Congenital Anomalies of the Upper Extremities

# 6

Sondra Elice Solomon

You came so nearly perfect from the hand of nature that this slightest possible defect, which we hesitate whether to term a defect or a beauty, shocks me as being the visible mark of earthly imperfection [1].

---

## Introduction

In most cultures physical perfection is the standard by which a person's competence, intelligence, and humanity are assessed [2–5]. Visible attributes that challenge physical perfection are not well tolerated by normal-appearing others [3]. When a person possesses a visible attribute that does not conform to a narrowly defined metric of appearance acceptability, the bearer of that negatively valued visible attribute may be at risk for social exclusion, prejudice, discrimination, and stigma by perceived normal-appearing others [6–8]. Furthermore, when the visible attribute in question is determined by genetic or medical factors, psychological well-being may be affected [9].

Early consensus in the psychological literature suggested that individuals with visible atypical body or facial attributes would always be at a social disadvantage, since in addition to managing their own appearance-related thoughts, feelings, and behaviors, they had to manage the reactions of normal-appearing others towards their appearance [10]. Contemporary research acknowledges the complex interactions of individual, social, and cultural factors that shape the experiences and psychological well-being of individuals with atypical visible features [8, 11]. Facial appearance has

been at the forefront of this research, and, at first blush, it is easy to understand why disruptions in facial appearance receive so much consideration. The face is a primary vehicle of human communication, and individuals make immediate judgments about others based on facial appearance. When facial integrity is disrupted social interaction is disrupted [7, 12]. However, hands and arms have salient cultural meaning as well. Hands and arms are essential for [1] interacting with and manipulating the physical world; [2] communicating with others; and [3] establishing and maintaining intimate physical contact with others. Like the face, hands and arms are difficult to conceal. Disruptions in the appearance of hands and arms have the potential to affect psychological well-being, yet there is limited research on the psychological functioning of individuals living with visible characteristics associated with congenital anomalies of the upper extremities (CAUE).

---

## Definitions

In an effort to promote psychological well-being among individuals living with CAUE it might be useful to reevaluate the words we use to describe the population. The language we use to describe the people we treat has the potential to foster a strong therapeutic alliance as we work towards promoting long-term positive adjustment and psychological well-being for our patients. It is suggested that those who serve individuals living with CAUE employ the terminology offered in the following section. When possible and appropriate these terms will be used throughout this chapter.

## Distinction

The term *distinction* will be used when referring to what the CAUE literature has characterized as aberrant, deformed, disfigured, defective, deficient, malformed, and abnormal attributes.

---

S.E. Solomon, Ph.D. (✉)

College of Arts and Sciences, Department of Psychological Sciences, University of Vermont, Dewey Hall, 346; 2 Colchester Avenue, Burlington, VT 05446, USA

College of Medicine, Department of Psychiatry, University of Vermont, Dewey Hall, 346; 2 Colchester Avenue, Burlington, VT 05446, USA  
e-mail: [Sondra.Solomon@uvm.edu](mailto:Sondra.Solomon@uvm.edu)

The term *distinction* is relatively benign and can be substituted for the pejorative and negative labels that describe the visible characteristics of CAUE that affect appearance. It is recognized that some authors prefer the term *physical difference* [11, 13]; however, *distinction* is a relatively neutral word and is an appropriate descriptor for a visibly and culturally devalued attribute related to appearance.

## Impairment and Disability

Most readers familiar with the genetics, rehabilitation, and disability literature will recognize the following definitions; however, it is useful to mention them again. *Impairment* is linked to a loss or a disruption of an anatomical structure or function and can be biologically determined or acquired via a disease process during a person's life, and *disability* is the consequence of the impairment and involves any restriction in the person's ability to perform an activity in the manner or within the range considered appropriate for individuals without the impairment [14–17]. The disability is the physical consequence of the impairment and is linked to how the impairment is manifested in the culture (i.e., the child with a congenital below the elbow anomaly has difficulty with motor function). The term *handicap* or *social handicap* should only be used when one considers how the person with the impairment is treated in the culture (i.e., the adult with a congenital below the elbow impairment is denied housing or employment due to processes that involve prejudice, exclusion, and discrimination).

## Stigma and Stigmatization

Erving Goffman [18] began a discourse spanning 50 years, transforming the way we examine how human beings manage the minute and salient differences between us. These differences place most people in two camps based on various personal characteristics and attributes. People can join and or be excluded from the two camps based on where they happen to be at the time (cultural context) and which attributes are valued at the time (temporal salience). Also, it may be possible for a person to be a member of both camps at the same time. Undesirable attributes may be fixed and unquestionable (e.g., congenital disorders, facial distinctions, excessive weight, cognitive deficits, old age, ethnicity, disability, diagnoses of severe and/or chronic psychopathology, perceived to be engaged in non-normative behavior, etc.).

Goffman defined *stigma* as a spoiled identity or a deeply discrediting characteristic which may arise from physical deformities, blemishes of individual character that are interpreted to reflect weakness, unnatural passions, dishonesty; and one's lineage [18]. Possession of the devalued attribute or distinction places the affected individual at a social disadvantage.

One early model to explain this disadvantaged social status suggests that stigma is a form of deviance that leads perceived normal-appearing others to judge individuals with the stigma as unworthy for participation in most social interchanges. They are viewed as incompetent, unpredictable, unreliable, or threatening [19]. This perception places the individual beyond the protection of a number of implicit norms that regulate social interaction. The disruptive impact of the distinction may be a function of how visible the distinction is to others, how much of the person's body is affected by the distinction, and how easily the distinction can be identified or seen by others [19, 20].

Researchers have been trying to understand and deconstruct the various processes contributing to devalued identities and subsequent spoiled interactions that devolve from possession of or contact with the undesirable attribute. Some have noted that it is difficult to identify a single defining feature of stigma and suggest that stigmatized people are believed to possess a feature, quality, or trait that portrays a social identity that is devalued in a particular social context [21]. In this view, stigma arises from one's membership in a group or category that is negatively valued in a specific situation (i.e., the adolescent with a below the elbow congenital anomaly is unable to participate in an activity in the same way that adolescents without the congenital anomaly).

*Stigmatization* may be conceptualized as a social process that seeks to reproduce inequality and exclusion [22–25]. There is an interaction between the environment and the individual with the distinction to recreate and perpetuate social and structural inequalities [25, 26]. Individuals with a devalued visible attribute may experience rejection, discrimination, and exclusion and these experiences have the potential to shape psychological, cognitive, and affective responses that affirm or impede healthy behaviors and psychological well-being [5].

## Visible Distinctions and Stigma

An accepted definition of a *visible distinction* is that the attribute in question represents a departure from a culturally defined norm which is difficult to conceal from others and as a result the attribute has the potential to shape interpersonal interaction with perceived normal-appearing others [27]. The attribute is perceived by others to be atypical, non-normative, and noticeable and excludes those attributes that are consistent with a body dysmorphic presentation [11, 27]. A *visible distinction* can have a powerful influence on the affected individual. A *visible distinction* is a social disability, since in addition to influencing the thoughts, feelings, and behavior of the person with the visible distinction, it is also likely to shape the behavior of other people towards the affected individual [28, 29]. Research suggests that the extent to which a visible distinction results in social disability

involves a complex interaction of social and individual factors [8, 11, 30]. We live in a culture that emphasizes physical perfection and individuals who possess visible attributes that are devalued occupy a special role in the culture and this role places them at a distinct advantage. The narrowly defined cultural appearance standard dictates who is accepted and who gets cast aside.

### **Stigma, Stigmatization, and Coping with Visible Distinctions**

The stigmatized person is diminished in the eyes of the observer and may experience a variety of stressors. A stressor is an event in which environmental or internal demands tax or exceed the adaptive resources of the individual [31]. Stigma can increase demands on the affected individual because perceived normal individuals may hold stereotyped expectancies about what stigmatized people are like, harbor prejudiced attitudes towards stigmatized people, and behave in a discriminatory manner towards stigmatized people [32, 33]. Psychological responses such as anger, anxiety, helplessness, resentment, and fear [3–5, 31, 34] may be experienced by the affected individual.

Visible distinctions are particularly stigmatizing because they remind the observer that the body is fragile, and depending on the etiology of the distinction may compel the observer to feel less compassion towards the individual with the distinction and to attribute more blame to them for having the distinction [35–37]. Children, adolescents, and adults living with CAUE frequently have visible attributes involving variations of limb formation, differentiation, duplication, overgrowth, and undergrowth; congenital constriction band syndrome; generalized skeletal irregularities; and comorbid facial irregularities [38]. Distinctions, such as those that can occur in CAUE, are particularly stigmatizing because the actual social identity—the attribute the individual possesses—does not meet society’s normative expectations of the attribute the individual should possess [26]. Social identity is flawed and the affected individual is presumed unable to fulfill the basic requirements of social interaction. Physical perfection is the gold standard for social inclusion. Social exclusion and subsequent threats to psychological well-being may be inevitable if the devalued attribute is visible and involves the hands and arms.

### **Psychological Research on Visible Distinctions Associated with CAUE**

Investigators have begun to explore how individuals adapt to a variety of stigmatizing attributes (e.g., diabetes, cancer, altered body appearance, HIV) [39–43]. An excellent review of the processes involved in managing visible distinctions

acknowledged that successful outcomes are linked to (1) the individual’s perception of the visible distinction; (2) their self-concept; (3) perceived and actual social support; (4) cultural contexts; (5) interpersonal encounters with others; (6) and the social skills they employ to manage difficult social encounters [11]. While this review was useful for a general understanding of psychological adjustment for those with visible distinctions (e.g., burns, dermatological disorders and cleft-palate) there was little to offer regarding those living with CAUE.

The broad spectrum of CAUE is rare but not entirely infrequent with a prevalence of 6.5–21.5 cases per 10,000 births [44]. They represent complex and variable pathologies with regard to the clinical severity of symptom presentation [45]. Some CAUE present in isolation and others present with associated systemic disorders and skeletal discrepancies [46]. Classification systems for the CAUE have been previously described [38, 46, 47]; however, the taxonomy endorsed by the International Federation of Societies for Surgery of the Hand is widely accepted [48, 49]. CAUE can be diagnosed in utero, at birth or during early childhood, and decisions regarding surgical intervention vary depending on the presentation of the specific genetic condition.

The Preferred Reporting Items for Systematic Reviews and Meta-analyses (PRISMA) [50] criteria were followed in an attempt to identify studies on psychological sequelae for individual living with visible distinctions related to CAUE. Articles published as of August 2013 in English using literature searches of Pubmed, Web of Science, and Psych Info were sought. Searches of the literature were conducted using the terms: upper extremity congenital anomalies, limb deficiencies, and hand and arm, in conjunction with one or more of the following key terms: psychosocial, adjustment, coping, well-being, quality of life (QOL), and appearance. It was difficult to identify empirical studies published during the past decade in which psychological well-being, coping, and adjustment to CAUE appearance-related concerns were the primary outcome variables.

The preponderance of the research, energy, and attention on CAUE has focused on neonates, children, adolescents, and families. Most studies have been concerned with the timing of the surgery during childhood, surgical intervention, or postsurgical satisfaction [51–54]; and, longer-term functional outcomes [55–60]. There are few studies on adults living with CAUE [61–65].

### **Children and Adolescents Living with a CAUE**

Some studies have demonstrated that living with a CAUE has an effect on the child’s and adolescent’s psychological well-being across several domains including self-esteem, internalizing behaviors (e.g., depression), and social interaction [66–70]. For example, one study of 66 children and

adolescents living with a CAUE fitted with a myoelectric prosthetic hand reported that there were higher levels of withdrawn behavior for all children and adolescents living with a CAUE compared to a normative sample, and, that females living with a CAUE reported lower social interaction competence: when compared to their male counterparts [66]. This finding is not surprising given the prevailing negative cultural attitudes towards visible physical distinctions which are particularly salient for females.

Recently, participation in day-to-day activities and QOL has received attention in the literature on successful outcomes for children and adolescents living with a CAUE. Participation and QOL can be viewed as proxy measures for psychological well-being. Participation is the extent to which an individual is involved in various life situations and may include, but not be limited, to the cultural context or attitudes of community members; family interest in recreation; and the affected individual's personal characteristics (e.g., gender and social competence) [71]. QOL refers to an individual's perceptions of their position in life within cultural and value systems in which they live and in relation to their goals and expectations. QOL, which has been used in lieu of psychological symptomology, can comprise physical, psychological, spiritual, environmental, and interpersonal domains [72].

Depending on the severity of the disorder, it may be presumed that children and adolescents with CAUE may be at risk for limited participation in social activities and report poor QOL and psychological well-being, yet the literature reports inconsistent findings. A recent narrative review of 15 cross-sectional studies of children and adolescents with congenital limb deficiencies noted that the literature lacks sufficient information to support or refute this presumption and further acknowledged that while full participation and enhanced QOL are considered the main goals in pediatric rehabilitation the literature provides limited empirical data on how children and adolescents with CAUE participate and how they view their QOL [71]. These authors also note that while some of the studies in their review used sound psychometric measures, most studies used small sample sizes, and employed descriptive, exploratory and cross-sectional research designs [71]. They also reported that direct comparison between the studies was difficult due to the wide age range in the study samples (2–20 years) the lack of knowledge regarding the heterogeneity of CAUE [71]. A 2012 qualitative study [73] of 42 children and adolescents between the ages of 8 and 20 years of age with unilateral congenital below the elbow deficiencies (UCBED) found the majority of respondents did not report limitations in self-care, school, or recreational activities. While older respondents reported difficulties with novel social encounters, they were attributed to restrictions placed on them by their school or work environment and not to appearance-related concerns [73].

The experience of living with a visible distinction associated with a CAUE during adolescence has not been thoroughly examined. Research on the social psychology of facial appearance has documented that conditions that threaten appearance may place the adolescent at risk for psychosocial and interpersonal challenges [74]. Studies examining the significance of visible distinctions on psychological well-being have emphasized a number of psychosocial challenges, including those related to social interaction [42], and the potential impact of negative self-perceptions on the development of the self-concept [75], and the ability to initiate and maintain romantic relationships [74]. A recent study acknowledged that poor psychological adjustment, specifically internalizing behaviors (e.g., depression) and poor health-related QOL were predicted by the adolescent's reports of perceived stigmatization (e.g., absence of friendly behavior, staring, hostile behavior) [76]. Yet another study reported positive adjustments to visible facial distinctions [77] and noted that protective factors (e.g., positive self-schemas, strong family ties and external social supports) could counteract appearance-related distress. While it is encouraging to report these findings, it is distressing that the question of whether an adolescent living with a CAUE is more or less likely to experience psychological distress during this developmental period remains unanswered. Perhaps CAUE-related visible distinctions may also result in similar outcomes.

### **Adults Living with a CAUE**

There are few studies on adults living with CAUE or on aging with CAUE. Case studies and reports on physical function are common [61–64]. One study commissioned by the Thalidomide Trust [61] reviewed the current health status and psychosocial sequelae of adults living with the consequences of Thalidomide in the United Kingdom. Of the 400 adults living with Thalidomide-related difficulties in the UK merely 12 men and 16 women participated in this study. The authors acknowledged these participants were married or had partners, many were employed reported good QOL and did not define themselves as disabled [61]. While these findings are encouraging it is difficult to determine if other adults would offer similar reports given the study's small and biased sample size. Furthermore, this study did not examine the appearance-related concerns related to living with a visible distinction.

### **Parental Coping and the Child with CAUE**

Parental coping and adjustment to the birth of a child with a CAUE is an emotional family event [78]. Parental adjustment to the distinction, and associated medical, financial, social, and emotional demands may place enormous stressors on the

family system [79, 80]. Parents face multiple challenges involving the management of grief-related emotions, finding an appropriate way to communicate with their children, and, making appropriate medical decisions [73]. Immediate- and longer-term factors contributing to the level of family distress may include but not necessarily be limited to (1) the extent and severity of the impairment and visibility of the distinction; (2) preexisting parental coping strategies; (3) the family's economic and psychological resources; (4) prevailing cultural attitudes towards the appearance of the child; and, (5) the developmental age of the child [81, 82].

Visible distinctions associated with CAUE may sometimes bias or otherwise impede a parent or caregiver's ability to effectively bond with their child [83, 84]. A successful transition through the first year of life characterized by bonding and parental affection and consistency in care are necessary conditions for the development of a child's sense of separate and valued self, and for the development of positive self-esteem [85, 86]. Researchers exploring family adjustment to the presence of a child with a visible distinction noted parents have reported heightened distress levels and that Parental psychological well-being prior to the birth of a child with a CAUE may be related to their child's long-term adjustment and psychological well-being [87]; however, it should be noted that findings are not consistent across studies due to inconsistencies in methodological approaches, small sample sizes, and scant longitudinal data. Such an approach may permit the development of integrated interventions within a biosocial medical model to improve functioning within this population.

---

## Directions for Future Research

The research on the psychosocial sequelae of individuals living with visible distinctions associated with CAUE is limited and findings are inconsistent. CAUE research energy and attention has centered on children, adolescents, and families. Data on the transition between adolescence to the early adult years are not evident. Data on adults coping with visible distinctions associated with CAUE are modest and few investigators have made coping with CAUE in across the lifespan a priority. For adults living with a CAUE case studies or the personal narrative within the context of overcoming adversity prevails. Perhaps there is a presumption that the adult with a CAUE would have few, if any, appearance-related concerns because child and adolescent issues have been resolved and the adult should have "gotten over it by now." Published studies are hindered by the lack of psychometrically validated measures and methodological approaches that were descriptive or qualitative. It is also noted that sufficient funding to support basic, clinical/translational research and, clinical intervention trials is limited. While there is a need for high-quality research in this area we should not be discouraged.

The open landscape offers an opportunity to develop a research agenda with an eye towards intervention.

We know from the extensive literature on psychological difficulties associated with facial appearance that the most common problems affected individuals encounter relate to negative self-perceptions, anticipatory anxiety regarding negative evaluations by others, and difficulties with social interactions [27]. Also, in contrast to early research examining the difficulties that individuals with visible facial distinctions encounter, investigators are devoting attention to the factors associated with adaptive coping strategies that affected individuals employ to manage a frequently hostile and unpredictable social landscape. The extensive literature on coping with stigmatizing attributes (e.g., obesity, HIV/AIDS) [25, 43, 88, 89] may provide some direction as well.

Coping has been defined as cognitive, emotional, and behavioral strategies that individuals employ to manage a variety of stressful experiences [31]. One coping model proposes two key responses: engagement and disengagement coping [90, 91]. Engagement coping can best be described by behaviors that engage with the stressful situation and/or by responses that help the individual to adapt to the stressful situation [43]. For example, the individual with a below the elbow anomaly may be confronted by persistent and unwelcome inquiries about his or her appearance. In response to these questions the affected individual may have at-the-ready a repertoire of responses to offer the curious observer. Disengagement coping involves responses that distance the individual from the stressor and includes avoidance, denial, and/or wishful thinking [43]. In this instance, the affected individual may avoid social encounters or engage in ruminative thoughts about his/her visible distinction.

Prior research also has demonstrated that the stigma associated with HIV poses various psychological challenges to people living with HIV, and that the consequences of stigma-related stressors on psychological well-being depend on the ability of affected individuals to employ engagement coping strategies [43, 92]. The stigma associated with CAUE appearance-related stigma may similarly pose psychological challenges to individuals living with visible distinctions associated with these anomalies yet little is known about these processes. It may be useful to employ stress and coping models to inform future research.

Researchers might examine the relationship between reports of appearance-related stigma; coping strategies used to manage the stigmatizing events, and associated psychological outcomes (e.g., depression, anxiety, anxiety sensitivity, resilience). What is the role of severity and visibility of the visible distinction? Are severity and visibility predictors of psychological difficulty? Are there risk or protective factors that may enhance positive outcomes? Are women, older adults, members of under-represented groups (e.g., African Americans, Latino/s, economically disadvantaged) at greater risk? What is

the role of, social support, family and cultural context in the management of CAUE appearance-related stigma?

Investigators should use normative groups of similarly aged individuals without a CAUE or compare findings to the reports of a first-degree relative (e.g., same gendered non-affected sibling). Reliable and valid instruments to measure coping, perceptions of appearance-related stigma, and psychological outcomes must be used. Longitudinal studies would also be beneficial.

These factors should be considered in future research protocols. Findings from this preliminary wave of research may inform appropriate interventions.

---

### **Some Closing Comments and a Personal Story: It's Not About Me**

The opportunity to write about the psychosocial aspects of living with a visible distinction associated with CAUE brought to mind my experience as a clinical psychologist, researcher, professor, and woman of color who lives with a genetic disorder and comorbid visible distinctions. Neurofibromatosis 1 (NF1), von Recklinghausen's disorder, or peripheral NF is one of several autosomal dominant neuro-cutaneous disorders caused by mutations of the gene on chromosome 17 (17q11.2) responsible for cell division [93]. Prevalence of NF1 is approximately 1 in 3,500 live births, and the disorder is highly random or variable regarding the clinical severity of symptom presentation [94, 95].

Clinical expressions of NF1 include café-au-lait spots, hamartomas (Lisch nodules), neurofibromas (Schwann cell tumors of four types: focal or diffuse cutaneous; subcutaneous; spinal; and, nodular or diffuse plexiform); optic gliomas; freckling in the axillary or inguinal regions; and distinctive bone lesions [93]. Common complications in individuals with NF1 are cognitive and learning disabilities [96]. While general intellectual functioning may be intact, identifiable and explicit cognitive deficits have been acknowledged among some affected individuals (e.g., perception, attention, executive functioning, language functions, learning disabilities, and visuo-spatial deficits) [97]. Surgical interventions to ameliorate or manage tumor growth have been reported in the literature. For example, surgical excision of plexiform neurofibromas of the face is complex and may require several medical interventions to debulk tumor growth; however, the cosmetic result is sometimes disappointing [98–100]. Also, individuals with NF1 are followed by various medical and mental health specialists (e.g., neurologists, neurosurgeons, ophthalmologists, orthopedic surgeons, reconstructive surgeons, genetic counselors, special educators, social workers, physical therapists, psychiatrists, neuropsychologists, psychologists, social workers) to manage symptoms, problems, or multiple impediments.

While most NF 1 tumors are benign, some individuals experience psychological distress as a result of the distinctive appearance associated with multiple visible tumors. Why mention NF 1 in a chapter devoted to CAUE? Individuals living with CAUE and NF1 may share some appearance-related concerns due to the visible distinctions associated with each disorder. I thought it would be useful to provide readers of this chapter with a first-hand account of what it is like to live with a visible distinction with an eye towards enriching clinical practice and research.

During my second year of doctoral training at the University of Vermont I was enrolled in a seminar in Community Clinical Psychology. On the first day of class we were asked to answer the following question: What is important to know about you? While not a fan of the “ice-breaker exercise” when it was my turn I complied and told my story. I said that I grew up in a housing project in New York City, in the north east Bronx. When I was a young girl, New York City housing projects were transitional housing for the upwardly mobile working class of the late 1950s, early 1960s. The Bronx River Housing Project was a diverse community of Europeans, African Americans, Latinos, South Americans, Pacific East Islanders, and Asians. Of course, we did not call ourselves by those names back then (we were Negro, Jew, Oriental, Irish, Italian, Greek, French, German, etc.). One day my father and mother announced to my sister and me that he had bought a home and we were going to move from the northeast Bronx to Riverdale. Riverdale was and remains an upscale residential community in the northwest Bronx. We were one of the first families of color to move to the area. To this day I do not know how my father was able to gather the financial resources to purchase a home for his family. At the time he worked for City of New York and earned \$75.00–\$100.00 a week. I told the class that my father inspired me and continues to inspire me. He never said these words to me explicitly, but the implicit message that my father's behavior modeled for me was that as long as you are alive you can do, shape, or change anything. As long as you have a goal, a dream, and a neuron firing in your skull you can achieve a vision. Your life condition does not matter. Your economic status, appearance privilege, weight, or age does not matter. As long as you can move and think you can shape a plan and implement that plan. I told the class that was the reason I decided to return to graduate school to become a psychologist when I was 41 years old. I told the class that the most important thing to know about me was that I was resilient.

Later that evening, a friend and fellow student called me. He said that he was baffled by the story I told in class, asked why I told that story, and wondered why I didn't talk about my NF. While I told the students in that seminar a story about me, what they wanted to hear was a story about my appearance. When people meet me they want to know: “What are

those things on your skin?” “Why do you look like that?” “What is wrong with you?”

Individuals with visible distinctions must answer these questions every day. Parents have to answer these questions for their children. These questions are part of the stressors that individuals with visible distinctions encounter. More often than not individuals with visible distinctions have to make it easier for others to engage in social interactions with them. The burden of initiating and maintaining the social encounter is on the shoulders of the individual with the visible distinction. Perceived normal-appearing others ask questions that reduce their anxiety or personal curiosity. The bearer of a visible distinction is frequently in the spotlight, on display and under public scrutiny.

When asked about my visible distinction I must be ready to provide an answer. Children *always* receive my full attention and compassion because children are curious, and it is good practice to let them know that individuals who do not look like them should not be feared or avoided. The reader should know that I have encountered well-meaning individuals who said, “Sondra, I can’t imagine how you do it.” Others have said, “I can’t imagine what I would do if I looked like you.” As I listen to the familiar refrain I imagine they are waiting for me to share some special magical life skill I possess to manage my visible distinction related to NF 1. I used to engage in lengthy conversation with people. I noticed that when they were sufficiently satisfied with my answer they would walk away. Now, when I am asked that question I respond with a smile and say, “Yes, I imagine you can’t.” This response is my attempt to ally with the person who is confused and anxious about my appearance. This response shifts the burden away from me and directs it towards the person who was compelled to break the social contract. The question is not about me at all but is about the anxiety, fragility, and vulnerability experienced by normal-appearing others when they encounter children, adolescents, and adults who do not look like them. This response is part of a number of engagement coping strategies that I employ to deflect the slings and arrows of outrageous fortune that are part and parcel of living in a culture that demands perfection.

## References

- Hawthorne N. *The birthmark*. Mosses from an Old Manse. New York: Wiley and Putnam; 1846.
- Sheridan J, Scior K. Attitudes towards people with intellectual disabilities: a comparison of young people from British South Asian and White British backgrounds. *Res Dev Disabil*. 2013;34(4):1240–7. PubMed PMID: WOS:000316532700016. English.
- Kurzban R, Leary MR. Evolutionary origins of stigmatization: the functions of social exclusion. *Psychol Bull*. 2001;127(2):187–208. PubMed PMID: ISI:000170928000001.
- MacDonald G, Leary MR. Why does social exclusion hurt? The relationship between social and physical pain. *Psychol Bull*. 2005;131(2):202–23. PubMed PMID: ISI:000227423200004.
- Richman LS, Leary MR. Reactions to discrimination, stigmatization, ostracism, and other forms of interpersonal rejection: a multivariate model. *Psychol Rev*. 2009;116(2):365–83. PubMed PMID: ISI:000264913000004.
- Harcourt D, Rumsey N, Paraskeva N. Appearance and body image. *Psychologist*. 2011;24(12):895–6. PubMed PMID: WOS:000297614100025. English.
- Rumsey N. The psychology of appearance: why health psychologists should do looks. *Psychol Health*. 2008;23:16. PubMed PMID: WOS:000260047300010. English.
- Rumsey N, Clarke A, White P, Wyn-Williams M, Garlick W. Altered body image: appearance-related concerns of people with visible disfigurement. *J Adv Nurs*. 2004;48(5):443–53. PubMed PMID: WOS:000225369600003. English.
- Ablon J. The nature of stigma and medical conditions. *Epilepsy Behav*. 2002;3(6):S2–9. PubMed PMID: WOS:000184307700002. English.
- Macgregor FC. *After plastic surgery: adaptation and adjustment*. New York: Praeger; 1979.
- Thompson A, Kent G. Adjusting to disfigurement: processes involved in dealing with being visibly different. *Clin Psychol Rev*. 2001;21(5):663–82. PubMed PMID: WOS:000169442100001. English.
- Wallace M, Harcourt D, Rumsey N. Experiences of healthcare provision in adolescents with an altered appearance. *Psychol Health*. 2008;23:270. PubMed PMID: WOS:000260047300633. English.
- Egan K, Harcourt D, Rumsey N, Appearance RC. A qualitative study of the experiences of people who identify themselves as having adjusted positively to a visible difference. *J Health Psychol*. 2011;16(5):739–49. PubMed PMID: WOS:000291676700005. English.
- Scheer J, Groce N. Impairment as a human constant—cross-cultural and historical perspectives on variation. *J Soc Issues*. 1988;44(1):23–37. PubMed PMID: WOS:A1988N062700003. English.
- Susman J. Disability, stigma and deviance. *Soc Sci Med*. 1994;38(1):15–22. PubMed PMID: WOS:A1994MK27000003. English.
- Zola IK. Self, identity and the naming question—reflections on the language of disability. *Soc Sci Med*. 1993;36(2):167–73. PubMed PMID: WOS:A1993KE81500009. English.
- Van Brakel WH, Anderson AM, Mutatkar RK, Bakirtzief Z, Nicholls PG, Raju MS, et al. The participation scale: measuring a key concept in public health. *Disabil Rehabil*. 2006;28(4):193–203. PubMed PMID: WOS:000235427800002. English.
- Goffman E. *Stigma: notes on the management of spoiled identity*. Englewood Cliffs, NJ: Prentice-Hall, Inc.; 1963. 147 p.
- Elliott GC, Ziegler HL, Altman BM, Scott DR. Understanding stigma—dimensions of deviance and coping. *Deviant Behav*. 1982;3(3):275–300. PubMed PMID: WOS:A1982NV59700005. English.
- Stafford MC, Scott RR. Stigma, deviance and social control. In: Ainlay SC, Becker GB, Coleman SC, editors. *The dilemma of difference: a multidisciplinary view of stigma*. New York: Plenum Press; 1986. p. 77–90.
- Crocker J, Major B, Steele C. Social stigma. In: Gilbert D, Fiske ST, Lindzey G, editors. *Handbook of social psychology*. 4th ed. Boston, MA: McGraw Hill; 1998.
- Castro A, Farmer P. Understanding and addressing AIDS-related stigma: from anthropological theory to clinical practice in Haiti. *Am J Public Health*. 2005;95(1):53–9.
- Link BG, Phelan JC. Conceptualizing stigma. *Annu Rev Sociol*. 2001;27:363–85.
- Malcolm A, Aggleton P, Bronfman M, Galvao J, Mane P, Verrall J. HIV-related stigmatization and discrimination: its forms and contexts. *Crit Public Health*. 1998;8(4):347–70.

25. Parker R, Aggleton P. HIV and AIDS-related stigma and discrimination: a conceptual framework and implications for action. *Soc Sci Med*. 2003;57(1):13–24.
26. Mill JE, Edwards N, Jackson RC, MacLean L, Chaw-Kant J. Stigmatization as a social control mechanism for persons living with HIV and AIDS. *Qual Health Res*. 2010;20(11):1469–83. PubMed PMID: WOS:000283250900005. English.
27. Rumsey N, Harcourt D. *The psychology of appearance*. New York, NY: Open University Press; 2005. 237 p.
28. Macgregor FC. Social, psychological and cultural dimensions of cosmetic and reconstructive plastic-surgery. *Aesthetic Plast Surg*. 1989;13(1):1–8. PubMed PMID: WOS:A1989T620100001. English.
29. Macgregor FC. Patient dissatisfaction with results of technically satisfactory surgery. *Aesthetic Plast Surg*. 1981;5(1):27–32. PubMed PMID: WOS:A1981LM55500003. English.
30. Harcourt D, Rumsey N. Psychology and visible difference. *Psychologist*. 2008;21(6):486–9. PubMed PMID: WOS:000256831200018. English.
31. Folkman S, Lazarus RS, Dunkelshetter C, Delongis A, Gruen RJ. Dynamics of a stressful encounter: cognitive appraisal, coping and encounter outcomes. *J Pers Soc Psychol*. 1986;50(5):992–1003. PubMed PMID: WOS:A1986C306700013. English.
32. Crocker J, Major B. Social stigma and self-esteem: the self-protective properties of stigma. *Psychol Rev*. 1989;96(4):608–30. PubMed PMID: ISI:A1989AU66600004.
33. Miller CT, Kaiser CR. A theoretical perspective on coping with stigma. *J Soc Issues*. 2001;57(1):73–92. PubMed PMID: ISI:000168242500005.
34. Folkman S, Greer S. Promoting psychological well-being in the face of serious illness: when theory, research and practice inform each other. *Psychooncology*. 2000;9(1):11–9. PubMed PMID: ISI:000085499100002.
35. Ben-Naim S, Aviv G, Hirschberger G. Strained interaction: evidence that interpersonal contact moderates the death-disability rejection link. *Rehabil Psychol*. 2008;53(4):464–70. PubMed PMID: WOS:000261518900005. English.
36. Hirschberger G. Terror management and attributions of blame to innocent victims: reconciling compassionate and defensive responses. *J Pers Soc Psychol*. 2006;91(5):832–44. PubMed PMID: WOS:000241548100003. English.
37. Hirschberger G, Florian V, Mikulincer M. Fear and compassion: a terror management analysis of emotional reactions to physical disability. *Rehabil Psychol*. 2005;50(3):246–57. PubMed PMID: WOS:000231741200009. English.
38. Kozin SH. Upper-extremity congenital anomalies. *J Bone Joint Surg Am*. 2003;85A(8):1564–76. PubMed PMID: WOS:000184552100021. English.
39. Schabert J, Browne JL, Mosely K, Speight J. Social stigma in diabetes a framework to understand a growing problem for an increasing epidemic. *Patient*. 2013;6(1):1–10. PubMed PMID: WOS:000315586900001. English.
40. Sterba KR, Zapka J, Gore EI, Ford ME, Ford DW, Thomas M, et al. Exploring dimensions of coping in advanced colorectal cancer: implications for patient-centered care. *J Psychosoc Oncol*. 2013;31(5):517–39. PubMed PMID: WOS:000324001300003. English.
41. Rumsey N. Body image and congenital conditions with visible differences. In: Cash TF, Pruzinsky T, editors. *Body image: a handbook of theory, research, and clinical practice*. London: The Guilford Press; 2002.
42. Rumsey N, Clarke A, White P, Wyn-Williams M, Garlick W. Altered body image: appearance-related concerns of people with visible disfigurement. *J Adv Nurs*. 2004;48(5):443–53. PubMed PMID: MEDLINE:15533082. English.
43. Varni SE, Miller CT, McCuin T, Solomon S. Disengagement and engagement coping with HIV/AIDS stigma and psychological well-being of people with HIV/AIDS. *J Soc Clin Psychol*. 2012;31(2):123–50. PubMed PMID: WOS:000300333400002. English.
44. Giele H, Giele C, Bower C, Allison M. The incidence and epidemiology of congenital upper limb anomalies: a total population study. *J Hand Surg*. 2001;26(4):628–34.
45. Ardon MS, Janssen WG, Hovius SE, Stam HJ, Selles RW. Low impact of congenital hand differences on health-related quality of life. *Arch Phys Med Rehabil*. 2012;93(2):351–7. PubMed PMID: WOS:000300338900026. English.
46. Aucourt J, Budzik JF, Manouvrier-Hanu S, Mezel A, Cotten A, Boutry N. Congenital malformations of the hand and forearm in children: what radiologists should know. *Semin Musculoskelet Radiol*. 2012;16(2):146–58. PubMed PMID: WOS:000304677600009. English.
47. Linder JA, Pincus DJ, Panthaki Z, Thaller SR. Congenital anomalies of the hand: an overview. *J Craniofac Surg*. 2009;20(4):999–1004. PubMed PMID: WOS:000268400400008. English.
48. Manske PR, Oberg KC. Classification and developmental biology of congenital anomalies of the hand and upper extremity. *J Bone Joint Surg Am*. 2009;91A:3–18. PubMed PMID: WOS:000267673900002. English.
49. Oberg KC, Feenstra JM, Manske PR, Tonkin MA. Developmental biology and classification of congenital anomalies of the hand and upper extremity. *J Hand Surg Am*. 2010;35A(12):2066–76. PubMed PMID: WOS:000285371300024. English.
50. Moher D, Liberati A, Tetzlaff J, Altman DG, Grp P. Preferred reporting items for systematic reviews and meta-analyses: the PRISMA statement. *J Clin Epidemiol*. 2009;62(10):1006–12. PubMed PMID: WOS:000270250500003. English.
51. Bellew M, Haworth J, Kay SP. Toe to hand transfer in children: ten year follow up of psychological aspects. *J Plast Reconstr Aesthet Surg*. 2011;64(6):766–75. PubMed PMID: WOS:000290562600020. English.
52. Vasluian E, de Jong IGM, Janssen WGM, Poelma MJ, van Wijk I, Reinders-Messelink HA, et al. Opinions of youngsters with congenital below-elbow deficiency, and those of their parents and professionals concerning prosthetic use and rehabilitation treatment. *PLoS One*. 2013;8(6):e67101. PubMed PMID: WOS:000321738400104. English.
53. Staines KG, Majzoub R, Thornby J, Netscher DT. Functional outcome for children with thumb aplasia undergoing pollicization. *Plast Reconstr Surg*. 2005;116(5):1314–23. PubMed PMID: WOS:000232421100017. English.
54. Hardwicke J, Khan MAA, Richards H, Warner RM, Lester R. Macrodactyly—options and outcomes. *J Hand Surg Eur*. 2013;38E(3):297–303. PubMed PMID: WOS:000317916800011. English.
55. Hadders-Algra M, Reinders-Messelink HA, Huizing K, van den Berg R, van der Sluis CK, Maathuis CGB. Use and functioning of the affected limb in children with unilateral congenital below-elbow deficiency during infancy and preschool age: a longitudinal observational multiple case study. *Early Hum Dev*. 2013;89(1):49–54. PubMed PMID: WOS:000314073900010. English.
56. Ekblom AG, Dahlin LB, Rosberg HE, Wiig M, Werner M, Arner M. Hand function in children with radial longitudinal deficiency. *BMC Musculoskelet Disord*. 2013;14:116. PubMed PMID: WOS:000318296200001. English.
57. Korkmaz M, Erbahceci F, Ulger O, Topuz S. Evaluation of functionality in acquired and congenital upper extremity child amputees. *Acta Orthop Traumatol Turc*. 2012;46(4):262–8. PubMed PMID: WOS:000309801300007. English.



58. Nemeč SF, Kasprian G, Brugger PC, Bettelheim D, Amann G, Nemeč U, et al. Abnormalities of the upper extremities on fetal magnetic resonance imaging. *Ultrasound Obstet Gynecol.* 2011;38(5):559–67. PubMed PMID: WOS:000297512300014. English.
59. Oberg KC, Harris TE, Wongworawat MD, Wood VE. Combined congenital radial and ulnar longitudinal deficiencies: report of 2 cases. *J Hand Surg Am.* 2009;34A(7):1298–302. PubMed PMID: WOS:000269414600019. English.
60. Huizing K, Reinders-Messelink H, Maathuis C, Hadders-Algra M, van der Sluis CK. Age at first prosthetic fitting and later functional outcome in children and young adults with unilateral congenital below-elbow deficiency: a cross-sectional study. *Prosthet Orthot Int.* 2010;34(2):166–74. PubMed PMID: WOS:000283117200005. English.
61. Bent N, Tennant A, Neumann V, Chamberlain MA. Living with thalidomide: health status and quality of life at 40 years. *Prosthet Orthot Int.* 2007;31(2):147–56. PubMed PMID: WOS:000248726400004. English.
62. Holtslag I, van Wijk I, Hartog H, van der Molen AM, van der Sluis C. Long-term functional outcome of patients with longitudinal radial deficiency: cross-sectional evaluation of function, activity and participation. *Disabil Rehabil.* 2013;35(16):1401–7. PubMed PMID: WOS:000321465200011. English.
63. Goh ESY, Li CM, Horsburgh S, Kasai Y, Kolomietz E, Morel CF. The Roberts syndrome/SC phocomelia spectrum—a case report of an adult with review of the literature. *Am J Med Genet A.* 2010;152A(2):472–8. PubMed PMID: WOS:000274508300034. English.
64. Cetik O, Uslu M, Cirpar M, Eksioğlu F. Experience with the surgical treatment of radial polydactyly in adults. *Ann Plast Surg.* 2005;55(4):363–6. PubMed PMID: WOS:000232282800006. English.
65. Eskandari MM, Oztuna V, Demirkan F. Late psychosocial effects of congenital hand anomaly. *Hand Surg.* 2004;9(2):257–9.
66. Hermansson L, Eliasson AC, Engstrom I. Psychosocial adjustment in Swedish children with upper-limb reduction deficiency and a myoelectric prosthetic hand. *Acta Paediatr.* 2005;94(4):479–88. PubMed PMID: WOS:000228451400018. English.
67. Varni JW, Rubinfeld LA, Talbot D, Setoguchi Y. Determinants of self-esteem in children with congenital acquired limb deficiencies. *J Dev Behav Pediatr.* 1989;10(1):13–6. PubMed PMID: WOS:A1989T156700003. English.
68. Varni JW, Setoguchi Y. Screening for behavioral and emotional problems in children and adolescents with congenital or acquired limb deficiencies. *Am J Dis Child.* 1992;146(1):103–7. PubMed PMID: WOS:A1992GY44600027. English.
69. Varni JW, Setoguchi Y. Effects of parental adjustment on the adaptation of children with congenital or acquired limb deficiencies. *J Dev Behav Pediatr.* 1993;14(1):13–20. PubMed PMID: WOS:A1993KK64300003. English.
70. Varni JW, Setoguchi Y. Perceived physical appearance and adjustment of adolescents with congenital/acquired limb deficiencies: a path-analytic model. *J Clin Child Psychol.* 1996;25(2):201–8. PubMed PMID: WOS:A1996UN35700009. English.
71. Michielsen A, Van Wijk I, Ketelaar M. Participation and quality of life in children and adolescents with congenital limb deficiencies: a narrative review. *Prosthet Orthot Int.* 2010;34(4):351–61. PubMed PMID: WOS:000284360400001. English.
72. Cohen JS, Biesecker BB. Quality of life in rare genetic conditions: a systematic review of the literature. *Am J Med Genet A.* 2010;152A(5):1136–56. PubMed PMID: WOS:000277739800008. English.
73. de Jong IGM, Reinders-Messelink HA, Janssen WGM, Poelma MJ, van Wijk I, van der Sluis CK. Mixed feelings of children and adolescents with unilateral congenital below elbow deficiency: an online focus group study. *PLoS One.* 2012;7(6):e37099. PubMed PMID: WOS:000305336800003. English.
74. Griffiths C, Williamson H, Rumsey N. The romantic experiences of adolescents with a visible difference: exploring concerns, protective factors and support needs. *J Health Psychol.* 2012;17(7):1053–64. PubMed PMID: WOS:000309354700015. English.
75. Gussy M, Kilpatrick N. The self-concept of adolescents with cleft lip and palate: a pilot study using a multidimensional/hierarchical measurement instrument. *Int J Paediatr Dent.* 2006;16(5):335–41. PubMed PMID: WOS:000239876300004. English.
76. Masnari O, Schiestl C, Rossler J, Gutlein SK, Neuhaus K, Weibel L, et al. Stigmatization predicts psychological adjustment and quality of life in children and adolescents with a facial difference. *J Pediatr Psychol.* 2013;38(2):162–72. PubMed PMID: WOS:000315417800007. English.
77. Feragen KB, Kvaalem IL, Rumsey N, Borge AIH. Adolescents with and without a facial difference: the role of friendships and social acceptance in perceptions of appearance and emotional resilience. *Body Image.* 2010;7(4):271–9. PubMed PMID: WOS:000284348500002. English.
78. Bradbury ET, Kay SPJ, Hewison J. The psychological impact of microvascular free toe transfer for children and their parents. *J Hand Surg Br Eur.* 1994;19B(6):689–95. PubMed PMID: WOS:A1994QJ56000003. English.
79. Fonseca A, Nazare B, Canavarró MC. Parental psychological distress and quality of life after a prenatal or postnatal diagnosis of congenital anomaly: a controlled comparison study with parents of healthy infants. *Disabil Health J.* 2012;5(2):67–74. PubMed PMID: WOS:000301868700002. English.
80. Fonseca A, Nazare B, Canavarró MC. Clinical determinants of parents' emotional reactions to the disclosure of a diagnosis of congenital anomaly. *J Obstet Gynecol Neonatal Nurs.* 2013;42(2):178–90. PubMed PMID: WOS:000316279500009. English.
81. Warfield ME, Krauss MW, Hauser-Cram P, Upshur CC, Shonkoff JP. Adaptation during early childhood among mothers of children with disabilities. *J Dev Behav Pediatr.* 1999;20(1):9–16. PubMed PMID: WOS:000078764900002. English.
82. Hauser-Cram P, Warfield ME, Shonkoff JP, Krauss IV MW. Results: predictors of functioning and change in children's development and parent well-being. *Monogr Soc Res Child Dev.* 2001;66(3):54–78.
83. Wallander JL, Varni JW, Babani L, Banis HT, Wilcox KT. Family resources as resistance factors for psychological maladjustment in chronically ill and handicapped-children. *J Pediatr Psychol.* 1989;14(2):157–73. PubMed PMID: WOS:A1989AC65200002. English.
84. Wallander JL, Varni JW. Effects of pediatric chronic physical disorders on child and family adjustment. *J Child Psychol Psychiatry.* 1998;39(1):29–46. PubMed PMID: WOS:000072047800003. English.
85. Beuf AH. Children coping with impaired appearance: social and psychological influences. *Gen Hosp Psychiatry.* 1990;6:294–301.
86. Hillbeuf A, Porter JDR. Children coping with impaired appearance—social and psychologic influences. *Gen Hosp Psychiatry.* 1984;6(4):294–301. PubMed PMID: WOS:A1984TM50000009. English.
87. Wiegner S, Donders J. Predictors of parental distress after congenital disabilities. *J Dev Behav Pediatr.* 2000;21(4):271–7. PubMed PMID: WOS:000088913700003. English.
88. Puhl RM, Brownell KD. Confronting and coping with weight stigma: an investigation of overweight and obese adults. *Obesity.* 2006;14(10):1802–15. PubMed PMID: WOS:000249605900017. English.
89. Myers A, Rosen JC. Obesity stigmatization and coping: relation to mental health symptoms, body image, and self-esteem. *Int J Obes.* 1999;23(3):221–30. PubMed PMID: WOS:000078796900001. English.

90. Compas BE, Connor-Smith JK, Saltzman H, Thomsen AH, Wadsworth ME. Coping with stress during childhood and adolescence: problems, progress, and potential in theory and research. *Psychol Bull.* 2001;127(1):87–127. PubMed PMID: ISI:000166843700005.
91. Connor-Smith JK, Compas BE, Wadsworth ME, Thomsen AH, Saltzman H. Responses to stress in adolescence: measurement of coping and involuntary stress responses. *J Consult Clin Psychol.* 2000;68(6):976–92.
92. Varni SE, Miller CT, Solomon SE. Sexual behavior as a function of stigma and coping with stigma among people with HIV/AIDS in rural New England. *AIDS Behav.* 2012;16(8):2330–9. PubMed PMID: WOS:000310316200024. English.
93. Evans D, Howard E, Giblin C, Clancy T, Spencer H, Huson S, et al. Birth incidence and prevalence of tumor-prone syndromes: estimates from a UK family genetic register service. *Am J Med Genet A.* 2010;152(2):327–32.
94. Lu-Emerson C, Plotkin S. The neurofibromatoses. Part 2: NF2 and schwannomatosis. *Rev Neurol Dis.* 2008;6(3):E81–6.
95. Lu-Emerson C, Plotkin S. The neurofibromatoses. Part 1: NF1. *Rev Neurol Dis.* 2009;6(2):E47.
96. Hyman SL, Shores A, North KN. The nature and frequency of cognitive deficits in children with neurofibromatosis type 1. *Neurology.* 2005;65(7):1037–44.
97. Van Es S, North K, McHugh K, De Silva M. MRI findings in children with neurofibromatosis type 1: a prospective study. *Pediatr Radiol.* 1996;26(7):478–87.
98. Chaudhry IA, Morales J, Shamsi FA, Al-Rashed W, Elzaridi E, Arat YO, et al. Orbitofacial neurofibromatosis: clinical characteristics and treatment outcome. *Eye.* 2012;26(4):583–92. PubMed PMID: WOS:000302938500015. English.
99. Lantieri L, Meningaud JP, Grimbert P, Bellivier F, Lefaucheur JP, Ortonne N, et al. Repair of the lower and middle parts of the face by composite tissue allotransplantation in a patient with massive plexiform neurofibroma: a 1-year follow-up study. *Lancet.* 2008;372(9639):639–45. PubMed PMID: WOS:000258622200027. English.
100. Miyawaki T, Billings B, Har-Shai Y, Agbenorku P, Kokuba E, Moreira-Gonzalez A, et al. Multicenter study of wound healing in neurofibromatosis and neurofibroma. *J Craniofac Surg.* 2007;18(5):1008–11. PubMed PMID: WOS:000249894600004. English.