Long-Term Functional Upper-Extremity Outcomes in Adults with Apert Syndrome

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Background: The goal of this study was to determine upper-extremity function and health-related quality of life in a cohort of adults with Apert syndrome.
Methods: Twenty-two adults with Apert syndrome completed the Disabilities of the Arm, Shoulder, and Hand survey; the 36-Item Short-Form Health Survey; and a semistructured interview. One surgeon administered the Jebsen Hand Function Test and measured sensation, joint motion, and strength.
Results: Median Disabilities of the Arm, Shoulder, and Hand score was 16.9, which indicated slightly greater disability than the population norm of 10.1. Median 36-Item Short-Form Health Survey scores were 54.5 for mental health and 57.0 for physical health—both more favorable than population norms. Total Jebsen Hand Function Test scores for dominant hand were 69.2 seconds for men and 64.7 seconds for women versus 37.8 seconds for population norms of both sexes. More complex syndactyly resulted in worse metacarpophalangeal joint motion but no significant difference in Disabilities of the Arm, Shoulder, and Hand; 36-Item Short-Form Health Survey; or other functional results. There was no difference in self-reported outcomes between patients with four ($n = 8$) versus five digits ($n = 14$) in each hand.
Conclusions: In this cohort of adults with Apert syndrome, self-reported assessment of disability was more favorable than measured functional data would suggest. Despite significant functional deficits, the participants in this study had adapted remarkably well. (*Plast. Reconstr. Surg.* 143: 1136, 2019.)

Apert syndrome is a rare condition characterized by craniosynostosis, midfacial hypoplasia, and bilateral syndactyly of the hands and feet.[1] Hand syndactyly is clinically divided into three types depending on the degree of involvement. In type I, the thumb is free and the central three digits are coalesced and attached to the fifth digit by a simple incomplete syndactyly (obstetrician's hand). In type II, the thumb is attached with a soft-tissue web, the central three digits exhibit more complex coalition, and the fifth digit has a complete and sometimes bony syndactyly (cupped hand). In type III, the thumb and all fingers are involved with complex bony coalition (rosebud hand). In Apert hands, the fingers are all short and stiff. There is usually no motion at the interphalangeal joints, and the metacarpophalangeal joints exhibit varying degrees of stiffness. The small finger is usually least affected. The thumb is broad and short, and deviates radially. In addition to the hand differences, many patients also have shoulder and elbow involvement, further limiting upper-extremity function.[2]

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Several staging algorithms and technical variations have been proposed for syndactyly release in Apert syndrome.[3–9] These articles focus on short-term clinical outcomes in children and lack solid functional outcomes data. Other studies have measured health-related quality of life in patients with Apert syndrome to assess cognitive development, psychosocial outcomes, intelligence, and emotional health.[10–15] In contrast, there are few long-term data on extremity function and health-related quality of life. The purpose of this study was to evaluate these parameters in adults with Apert syndrome.

PATIENTS AND METHODS

Medical records at Boston Children's Hospital and a publicly available database (accessible at www.Apert.org) were queried to identify adults aged 18 to 65 years with Apert syndrome. Serial molds, records, pictures, and radiographs were available for the patients treated by the senior author (J.U.) (Fig. 1). Mortality status was checked against a public database (www.ancestry.org). Subjects were recruited by means of mail or e-mail and phone calls. Recruited subjects all had sufficient neurocognitive ability to read, write, and follow the appropriate directions for the measured tests and surveys.

Surveys and Interview

After obtaining written informed consent, researchers administered two self-report questionnaires: the 36-Item Short-Form Health Survey version 2, and the Disabilities of the Arm, Shoulder, and Hand outcome measure. The 36-Item Short-Form Health Survey is a 36-item instrument that assesses overall health[16] separated into two sections: mental and physical health. The Disabilities of the Arm, Shoulder, and Hand questionnaire is a 30-item instrument that measures disability in adult patients with upper-extremity conditions.[17]

Participants were also interviewed by a research assistant to discuss daily activities (e.g., school, work, volunteering), highest level of education, current living situation, and current social situation.

Functional Assessments

All assessments were conducted by a hand surgeon (A.H.T.) who provides surgical treatment for Apert syndrome, but who had not treated any of the participants. This surgeon assessed syndactyly type and recorded hand dominance. The number of digits present on each hand was recorded. For each digit, two-point discrimination was measured with a discriminator, and range of motion of the metacarpophalangeal joint was measured with a goniometer (individuals with Apert syndrome lack interphalangeal joint motion). For each hand, lateral (side-to-side) and chuck (tripod) pinch strength were measured with a pinch meter. Lastly, the Jebsen Hand Function Test, a timed test of hand function that asks the participants to perform seven subtasks,[18] was administered to all participants.

Ethics Statement

This study was approved by the Institutional Review Board at Boston Children's Hospital (IRB-P00018965). Following study completion, each subject was provided remuneration for their time and travel, and a copy of their individual results.

Statistical Analysis

Following double data entry into REDCap electronic data capture tools, quantitative data were analyzed using SAS software, Version 9.4 (SAS Institute, Inc., Cary, N.C.).

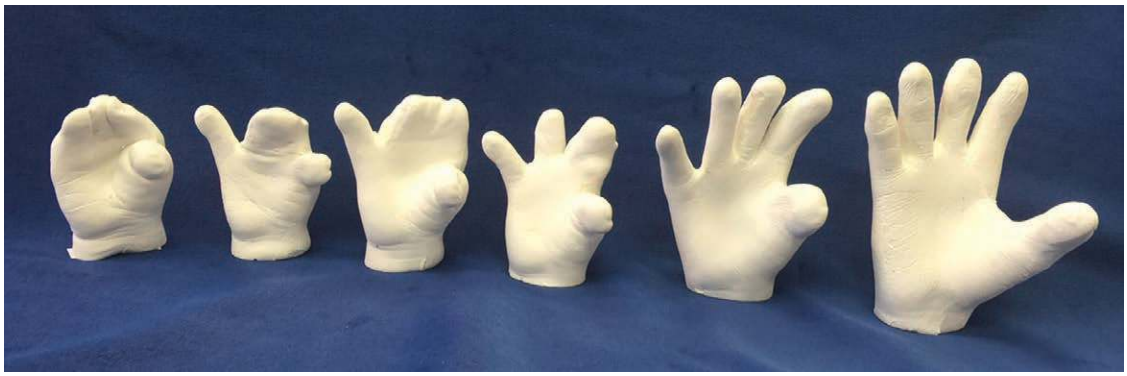

Fig. 1. Serial molds of one of the patients in this study treated by the senior surgeon (J.U.). A type II Apert hand is shown from infancy to adolescence demonstrating typical treatment progression and final outcome.

Table 1. Self-Reported Health Outcomes

Instrument	Median (IQR)	Best Possible Score	Population Norm (Average)
SF-36 Physical Health	57.0 (46.0–59.0)	100	50
SF-36 Mental Health	54.5 (43.0–61.0)	100	50
DASH	16.9 (4.17–24.2)	0	10.1

IQR, interquartile range; SF-36, 36-Item Short-Form Health Survey; DASH, Disabilities of the Arm, Shoulder, and Hand questionnaire.

RESULTS

From July of 2016 to November of 2017, 22 adults with Apert syndrome completed the study. Average participant age was 27 years (range, 18 to 43 years). Most were male (55 percent), Caucasian (91 percent), and right-hand dominant (64 percent). Twenty-seven percent had type I, 32 percent had type II, and 41 percent had type III syndactyly. Fourteen subjects had five digits reconstructed on both hands. Of the eight individuals (36 percent) who had four-digit reconstruction (both hands), two had type II and six had type III hands. All participants had been surgically treated for syndactyly and most had had other secondary procedures performed. The full surgical history of each participant was not available.

Self-Reported Health Outcomes

High scores on the 36-Item Short-Form Health Survey Physical Health and Mental Health subscores are more favorable and indicate better health for each category. The population norm score for each category of the 36-Item Short-Form Health Survey is 50; the best score is 100 and the worst score is 0. Median participant scores were 57.0 and 54.5 for the physical and mental health, respectively, and thus more favorable than population norms (Table 1).[16]

High scores on the Disabilities of the Arm, Shoulder, and Hand questionnaire indicate greater disability and are less favorable. The best possible score on the Disabilities of the Arm, Shoulder, and Hand questionnaire is 0, indicating no disability, whereas the worst possible score is 100, indicating total disability. Participants scored an average of 16.9 on this instrument, which was slightly higher than the population norm of 10.1,[19] indicating more disability and a less favorable score. There was no significant difference in self-reported outcomes between subjects who had four-digit reconstruction versus five-digit reconstruction.

Functional Outcomes

Patients with Apert syndrome typically lack active motion at the interphalangeal joints (as did all the subjects), so the total active motion of the digit is equivalent to the active motion of the metacarpophalangeal joint. The average maximum active extension of the metacarpophalangeal joint was 0.2 ± 1.5 degrees for the thumb, 2.2 ± 10.0 degrees for the index finger, 6.8 ± 12.8 degrees for the long finger, 1.8 ± 6.6 degrees for the ring finger, and 0.3 ± 2.5 degrees for the fifth finger. The average total active motion of each digit is outlined in Figure 2.

Average total active motion was stratified for each digit by hand type (Table 2). There was

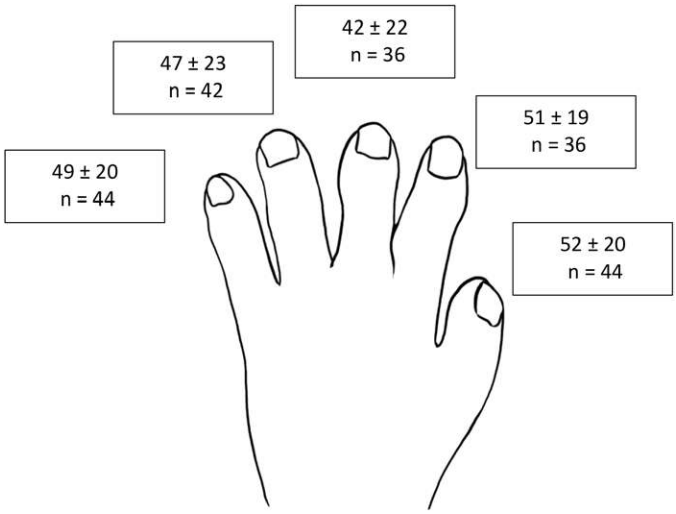

Fig. 2. Average metacarpophalangeal joint range of motion. Note that a few subjects had digital amputations.

Table 2. Average Total Range of Motion by Hand Type

	Type I	Type II	Type III	ANOVA *p*	Tukey Test
No.	6	7	9		
TAM by digit (degrees)					
Thumb	60.8 ± 18.7	53.9 ± 16.1	45.6 ± 16.1	NS	
Index	64.0 ± 17.2	50.4 ± 13.0	20.8 ± 22.5	<0.001	1–3, 2–3
Long	59.2 ± 22.5	30.0 ± 21.6	21.7 ± 17.3	0.007	1–2, 1–3
Ring	60.0 ± 24.2	49.6 ± 17.3	31.1 ± 20.3	0.040	1–3
Fifth	55.8 ± 15.9	52.9 ± 16.0	42.2 ± 21.6	NS	

ANOVA, analysis of variance; TAM, total active motion; NS, not significant ($p > 0.05$).

a notable trend indicating average total active motion was less for each digit with increasing complexity of the syndactyly. Patients with type I hands had higher average total active motion for each digit than type II, and patients with type II hands had higher total active motion than those with type III. This difference was statistically significant (by analysis of variance) only for the index, long, and ring fingers. The Tukey test was performed to control for type I errors, and showed which syndactyly types are different from each other.

Moving two-point discrimination was measured and reported by digit (Fig. 3). There did not appear to be any clinically significant difference in the sensibility of the measured digits, and values were inline with normal.

Average chuck (tripod) and lateral (side-to-side) pinch strengths were lower than population norms.[20] Average lateral pinch strength was 61 percent of norms in men and 90 percent of norms in women. Average chuck pinch strength was 48 percent of norms in men and 78 percent of norms in women. A one-sample *t* test showed that the difference in pinch strength in men was statistically significant. Among female subjects, however, only difference in chuck pinch was statistically significant (Table 3). There was no significant difference in pinch strength between syndactyly types.

Average times for each task on the Jebsen Hand Function Test were compared to normative values by calculating percentage difference and conducting a one-sample *t* test (Tables 4 and 5). Other than short sentence writing, the time taken by study subjects for all tasks was significantly longer than population norms—regardless of hand dominance or sex. Both sexes were slowest at stacking checkers and lifting large heavy objects. In comparing Jebsen subtask performance between hand types, there was no statistically significant difference in any of the measures except for page turning: those with type II hands appeared to perform this task more slowly by approximately 2 seconds.

We did not assess differences in functional outcomes between those who had four-digit versus five-digit reconstructions because controlling for hand dominance and sex diluted the sample sizes significantly and disallowed meaningful comparisons.

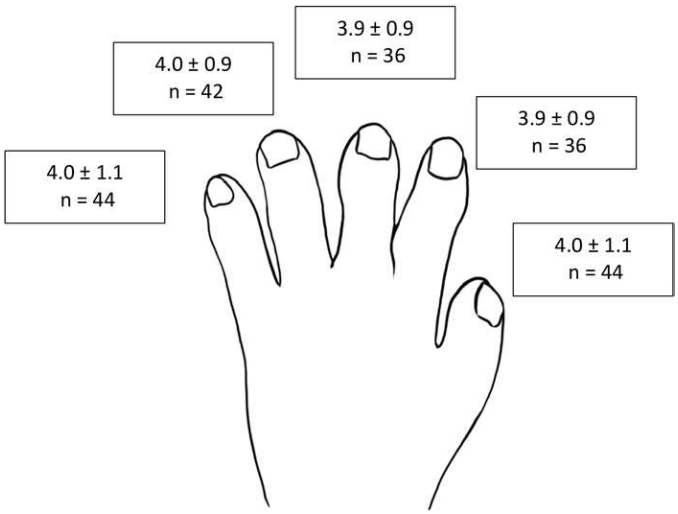

Fig. 3. Average moving two-point discrimination (in millimeters).

Table 3. Pinch Strength Values and Population Norms

	Male (n = 12)			Female (n = 10)		
	Study Group (kg)	Norm (kg)	p	Study Group (kg)	Norm (kg)	p
Lateral						
Right	6.8 ± 2.2	11.1 ± 2.1	<0.001	6.70 ± 3.01	7.4 ± 1.4	NS
Left	7.2 ± 2.5	10.7 ± 2.1	<0.001	6.36 ± 3.13	7.0 ± 1.4	NS
Chuck						
Right	5.1 ± 2.1	10.6 ± 2.3	<0.001	5.75 ± 2.09	7.4 ± 1.7	0.034
Left	4.7 ± 2.1	10.5 ± 2.4	<0.001	5.45 ± 1.71	7.1 ± 1.6	0.014

NS, not significant ($p > 0.05$).

Table 4. Jebsen Hand Function Test Scores for Male Participants (n = 12)

	Dominant (sec)				Nondominant (sec)			
	Study Group	Norm	Percent Difference	p	Study Group	Norm	Percent Difference	p
Short sentence	16.5 ± 6.0	12.2 ± 3.5	35	0.029	28.7 ± 7.9	32.3 ± 11.8	−11*	NS
Page turning	7.0 ± 1.7	4.0 ± 0.9	75	<0.001	6.9 ± 2.0	4.5 ± 0.9	53	0.002
Lifting small object	12.0 ± 4.4	5.9 ± 1.0	103	<0.001	13.0 ± 5.1	6.2 ± 0.9	110	<0.001
Simulated feeding	14.4 ± 7.2	6.4 ± 0.9	125	0.003	14.8 ± 5.2	7.9 ± 1.3	87	<0.001
Stacking checkers	7.9 ± 2.7	3.3 ± 0.7	139	<0.001	9.8 ± 4.4	3.8 ± 0.6	158	<0.001
Lifting large, light	5.8 ± 2.3	3.0 ± 0.4	93	0.001	7.0 ± 5.6	3.2 ± 0.6	119	0.039
Lifting large, heavy	5.6 ± 1.9	3.0 ± 0.5	87	<0.001	7.1 ± 3.6	3.1 ± 0.4	129	0.002
Total	69.2	37.8	83		87.3	61.0	61	

NS, not significant ($p > 0.05$).
*Represents a percentage decrease compared with normative value.

Table 5. Jebsen Hand Function Test Scores for Female Participants (n = 10)

	Dominant				Nondominant			
	Study Group	Dominant Norm	Percent Difference	p	Study Group	Nondominant Norm	Percent Difference	p
Short sentence	16.8 ± 5.2	11.7 ± 2.1	44	0.013	32.1 ± 8.4	30.2 ± 8.6	6	NS
Page turning	6.8 ± 2.2	4.3 ± 1.4	58	0.006	7.4 ± 2.4	4.8 ± 1.1	54	0.008
Lifting small object	10.0 ± 1.4	5.5 ± 0.8	82	<0.001	11.2 ± 2.3	6.0 ± 1.0	87	<0.001
Simulated feeding	11.3 ± 3.0	6.7 ± 1.1	69	0.001	14.2 ± 5.5	8.0 ± 1.6	78	0.006
Stacking checkers	6.9 ± 1.3	3.3 ± 0.6	109	<0.001	8.0 ± 1.4	3.8 ± 0.7	111	<0.001
Lifting large, light	6.0 ± 1.6	3.1 ± 0.5	94	0.000	6.1 ± 1.9	3.3 ± 0.6	85	0.001
Lifting large, heavy	6.9 ± 2.9	3.2 ± 0.5	116	0.003	8.7 ± 6.5	3.3 ± 0.5	164	0.026
Total	64.7	37.8	71		87.7	58.9	49	

NS, not significant ($p > 0.05$).

Interview Results

Education

Most subjects (17 of 22) had completed high school, four were still in high school or a technical school, and one was completing a post–high school program. Four had completed a 4-year undergraduate program, three were enrolled, three were planning to enroll, and one had enrolled but left without completion. One subject was in a graduate program, and two had started graduate programs but left without completion.

Experience

All subjects reported employment history, paid and voluntary, in a variety of fields. A few subjects specifically mentioned activities that help others with similar disabilities. Two subjects reported periods of unemployment and difficulty securing jobs. Eight subjects had driver's licenses (although not all drove), and four were in the process of getting a driver's license.

Lifestyle and Social Interactions

Subjects reported a range of living situations, from living with immediate family (n = 17), with a spouse (n = 1), or alone (n = 4). Most subjects reported completing household activities, such as cleaning, laundry, and pet care. Most participated in athletic activities, including swimming, tennis, surfing, and team sports such as basketball and baseball. Several competed in the Special Olympics.

Subjects reported a range of social interactions, including those with immediate and extended family and friends. Several subjects had dated and one was currently dating. One subject was married and two subjects had children. Several reported social interactions with others with Apert syndrome. A few subjects described social anxiety, and one noted a lack of friends.

DISCUSSION

This study offers novel data on long-term functional upper-extremity and health-related quality of life outcomes in adults with Apert syndrome. In our study population, self-reported health-related quality of life outcomes were more favorable than functional measures alone would suggest. Average total Jebsen Hand Function Test times were significantly slower than population norms. Similarly, total active motion and pinch strength measures were very low, mostly because of the lack of interphalangeal joint motion in all digits. Nevertheless, despite the unfavorable measured functional data, average total scores for the 36-Item Short-Form Health Survey were more favorable than population norms, and average Disabilities of the Arm, Shoulder, and Hand questionnaire score indicated a relatively low level of perceived disability. Furthermore, the qualitative data collected from semistructured interviews suggest a high level of functioning among these adult patients with Apert syndrome.

Our findings in these adults mirror the findings of others who compared functional measures with self-reported health-related quality of life outcomes in adults with congenital hand differences. Goldfarb and colleagues studied 21 adult patients (25 wrists) who were treated with centralization for radial longitudinal deficiency. They found a 62 percent increase over population norms in the speed of performing the Jebsen Hand Function Test, but the Disabilities of the Arm, Shoulder, and Hand questionnaire only showed mild disability of 18. They concluded that hand function remained markedly abnormal in these patients, whereas upper-extremity disability was mild.[21] Stutz and colleagues assessed long-term outcomes after radial polydactyly correction in 43 thumbs. The treated thumbs had significantly weaker pinch strength than the unaffected thumbs of the same patients; however, the average Disabilities of the Arm, Shoulder, and Hand questionnaire score was only 4.5.[22] The same group also found similar results in a different cohort of patients who were treated with on-top–plasty for radial polydactyly construction.[23] Pinch strengths were below the 50th percentile of age-matched norms, and grip strength was just above the 50th percentile, but the average Disabilities of the Arm, Shoulder, and Hand questionnaire score was 4.3. Similar findings about an apparent dissociation between measured function and self-reported upper-extremity disability in adult patients with congenital hand differences has been confirmed by others as well.[24,25]

Nevertheless, polydactyly and radial longitudinal deficiency are often unilateral, and functional deficits in one limb can be overcome by a normal contralateral limb, thus leading to less disability. Such is not the case in Apert syndrome, where the complex syndactyly is always bilateral and enantiomorphic. Furthermore, although elbow and shoulder function was not directly measured in this study, many Apert syndrome patients have more proximal upper-extremity deficits, including radioulnar synostosis, humeral-ulnar synostosis (approximately 10 percent), and elbow and shoulder stiffness (that worsens as they age), thus further compromising function.[2] Nevertheless, these functional issues translate to only mild disability, with an average Disabilities of the Arm, Shoulder, and Hand questionnaire score of 16.9 in this study. Although this value is 6.8 points higher than the population norm, it is less than the minimum clinically important difference (the smallest change in an outcome measure that is perceived to be clinically important) for the Disabilities of the Arm, Shoulder, and Hand questionnaire score of 15.

The findings outlined above are not necessarily surprising to experienced clinicians who work with patients with congenital upper-extremity differences. Despite obvious significant functional deficits, patients often do well and can lead rich, extraordinary lives. Although there can be many reasons for this observation, one hypothesis is that patients born with these conditions are better able to adapt to their deficits during early life—an advantage not seen in older individuals with acquired hand conditions. This hypothesis advocates for completing functional upper-extremity operations early in childhood to take advantage of brain plasticity and adaptability.

Individuals with Apert syndrome exhibit symphalangism of the digits; thus, their only functional joints are the metacarpophalangeal joints. Traditionally, the fifth digit is considered the "best" digit in patients with Apert syndrome. It is least involved in the syndactyly, and its radiographic appearance tends to be more normal

than the others. However, our study shows that average metacarpophalangeal joint motion in the fifth digit in these patients was not significantly (clinically) different than in the other digits. The standard deviations for metacarpophalangeal joint measurements were quite wide. Indeed, several patients had multiple digits with less than 5 degrees of motion at the metacarpophalangeal joint. The surgeon who administered the functional tests observed that most patients had one or two "good" fingers that were used for opposition against the thumb. These fingers had the greatest metacarpophalangeal joint motion and provided the main mode of pinch function.

Metacarpophalangeal joint range of motion decreased with increasing complexity of the syndactyly. Patients with type I hands had better range of motion in all digits than patients with type II hands, and type III patients were the most deficient. Although this observation confirms the finding that increasing initial complexity leads to worse range of motion, there was no difference seen in the other functional measures or the self-reported outcomes. In other words, although those with worse hand involvement may have less range of motion, they do not experience any difference in timed tasks or upper-extremity disability. It is possible that most of the useful function is between the thumb and one (or two) other digit(s); thus, once the syndactylies are released, useful function would be similar among different hand types (the observation outlined in the previous paragraph supports this hypothesis). Regardless, the findings outlined are important when counseling parents of children with Apert syndrome who are concerned about long-term function. That the presence of four (versus five) digits did not appear to influence self-reported outcomes is also worth consideration in more difficult cases.

Observations from the timed Jebsen Hand Function Test tasks indicated significant deficits in stacking of checkers and lifting of large, heavy objects as compared to norms. In Apert syndrome patients, short fingers and the lack of distal digital motion inhibit volumetric grip and limit large object grasp. This is one of the reasons we did not measure grip—because individuals cannot wrap their hands around the dynamometer. Slow performance in stacking checkers is also likely attributable to poor digital motion. Stacking checkers involves careful balancing, which in turn requires proprioceptive feedback of "fast fibers," which are typically well-represented in the small joints of the hand. Small movements at the metacarpophalangeal joints translate into large movements at the fingertip. Without fine control more distally near the fingertip, careful balancing of small objects can be difficult, which explains these results.

Apert syndrome also affects the craniofacial skeleton and can cause abnormal head shape and midface hypoplasia. Given the social stigma associated with the craniofacial anomalies, it was crucial to provide additional health measures in this study that could help further elucidate health-related quality of life and social integration. The subjects in this study scored more favorably than population norms in both mental and physical health aspects of the 36-Item Short-Form Health Survey. The qualitative interviews further substantiated these results and showed acceptable social/societal integration. All participants had at least completed high school and had been employed; many of them had relationships and participated in social activities such as sports and other group endeavors. Experience and social integration in our series mirrored other studies' results.[6,26,27] One possible topic for future study would be to assess which of the major manifestations of this condition—craniofacial or extremities—have a greater effect on health-related quality of life, function, and social integration.

A major limitation of this study is the small sample size, which limits the power of statistical tests and generalizability of the results. Apert syndrome is rare, and recruitment efforts from a pediatric facility are limited because most participants are no longer patients. Other limits on generalizability of these data include social support and neurocognitive status. Most adults who were successfully enrolled received assistance in coordination and/or travel from family member(s). Patients with neurocognitive delay could not participate in the study, thus deeming a potentially nontrivial portion of this patient population ineligible, and potentially introducing selection bias. The prevalence of neurocognitive delay in this sampled population of patients is unknown, and we were unable to estimate how many patients may have not been successfully recruited as a result. When counseling parents about long-term upper-extremity function and disability, clinicians should indicate that the outcomes outlined herein were observed in patients with sufficient neurocognitive ability to complete surveys and participate in functional tests.

Another limitation of this study was that patients were treated by different (often multiple) surgeons at different (often multiple)

institutions. Although all of them had undergone syndactyly release procedures, a few had undergone additional procedures such as osteotomies and digital lengthening procedures. Lastly, there was no control in terms of the degree of involvement—as even within certain types of syndactyly, there can be great variation in bony and soft-tissue anatomy.[6] Nevertheless, to standardize these patients and/or restrict the study population based on certain metrics would severely limit the study population. Furthermore, this study did not aim to assess outcomes of syndactyly release or other procedures in these patients, but rather to provide a snapshot of functional outcomes and disability in an adult cohort of Apert patients who had undergone corrective procedures in the past.

This study offers novel data on long-term functional upper-extremity and health-related quality-of-life outcomes in adults with Apert syndrome. Self-reported health-related quality-of-life outcomes were more favorable than functional measures alone would suggest. The participants in this study showed remarkable social integration and physical and emotional resilience despite significant functional and social impediments. Even though functional measures showed less motion, less strength, and less speed than population norms, these measured deficits did not translate to functional disability as perceived by the patients themselves. Treatment protocols have evolved, and it is possible that future cohorts will display better functional outcomes and appearance and, perhaps, even less disability.

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PSYCHOSOCIAL INSIGHTS

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Over the past several decades, a number of plastic surgery centers around the world have investigated the psychosocial impact of being born with a congenital condition that affects physical functioning and/or appearance. The commonly held belief among laypersons is that children and adolescents who are disfigured suffer greatly with body image dissatisfaction, low self-esteem, depression, and social anxiety. While that is the unfortunate experience for a minority of individuals born with congenital conditions, most studies have suggested that individuals born with conditions that affect their appearance report a wide range of reactions, and importantly, many show few differences from their unaffected peers. The consensus belief among many experts in the field is that individuals born with congenital conditions are at increased risk for psychosocial issues, but these untoward outcomes are not universal.

Far less research has been conducted on the functional and psychosocial outcomes of children born with these conditions who are now adults. There are several reasons for this. First, even among centers that specialize in treating children with craniofacial conditions and related disorders, the rare nature of the conditions results in relatively small clinical populations over time. Second, as these children move through adolescence and early adulthood, many are lost to follow-up. Thus, the ability of investigators to locate them and engage them in clinical research studies is challenged.

The study by Taghinia and colleagues provides important information on the functional and psychosocial status of a sample of 22 young to middle-age adults born with Apert syndrome. Compared to population norms, these individuals, on average, show mild to moderate deficits in hand functioning. However, their self-report of their disability was more positive than seen on those objective measures. Similarly, as assessed by the "gold standard" measure of health-related quality of life (the Short-Form Health Survey-36), the sample had median scores on the physical and mental health composite scales that were more positive than population norms, suggesting higher levels of quality of life.

These findings highlight several important issues. First, as seen in studies of the psychosocial functioning of children and adolescents with craniofacial conditions,

there are few differences in the functioning of affected and nonaffected individuals when examined at the group level. On the individual level, however, the psychosocial reaction may be quite different. As is often seen clinically, some children with mild to moderate levels of disfigurement present with significant psychosocial and behavioral issues. In contrast, others with profound disfigurement will often display relatively little psychosocial distress. In recent years, the pediatric psychology literature has increasingly focused on the psychological construct of resiliency as a way to understand how some children and families respond incredibly well to the stress of a severe craniofacial condition or other chronic health issues. Measures of the construct are recommended for future studies of both pediatric and adult craniofacial populations.

Second, relatively few studies in this area have combined both functional and psychosocial measures of outcome. This is a particular strength of this study and is encouraged in future work as well. The results of this investigation also highlight the observation that there is often a disconnect between a patient's and a provider's assessment of outcome of a given treatment. Adults with Apert syndrome in the present study self-reported lower levels of disability than anticipated from the functional assessments. This is an important reminder for researchers and clinicians that the patient's perspective of his or her illness may be more meaningful to the patient that our medical assessment or opinion. This may be an example of resiliency at work—that even in the face of objective measures of functional limitations, individuals find a way to endure through the challenges of daily life but also experience a level of quality of life not all that different from their family and friends.

Finally, results of this study may have implications for the growing field of vascularized composite allotransplantation. As the field continues to develop, there needs to be additional consideration of the level of physical and psychosocial functioning that makes an individual appropriate for a vascularized composite allotransplantation procedure. In cases where an individual has been born without a hand, or has lost one or more hands following traumatic injury or illness, the ability to engage in activities of daily living and secure meaningful employment will likely influence both the patient's and transplant team's thoughts about the appropriateness of hand transplantation. Similarly, a patient's successful use or failure with a prosthetic hand(s) will be an important part of the medical decision-making process. While the present study suggests that, as a group, adults with Apert syndrome are functioning well, it may be that an individual with the syndrome, and who is struggling physically and psychologically, will present for a hand transplant procedure. This will give those who work in the field of vascularized composite allotransplantation additional opportunity to think about the important issues of physical and psychosocial functioning in making a determination about appropriateness for a transplant procedure.

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