

**THE UNIVERSITY CENTER FOR EXCELLENCE IN  
DEVELOPMENTAL DISABILITIES**

**DATA REPORT:**

**Service Provision for Children Affected by Cornelia de Lange  
Syndrome**

**February 2008**

**University of Connecticut  
A.J. Papanikou Center  
for Excellence in Developmental Disabilities Education, Research, and Service  
UConn Health Center – MC 6222  
263 Farmington Ave.  
Farmington, CT 06030**

# THE UNIVERSITY CENTER FOR EXCELLENCE IN DEVELOPMENTAL DISABILITIES

## DATA REPORT: Service Provision for Children Affected by Cornelia de Lange Syndrome

February 2008

### **INTRODUCTION**

This study focuses on families that are affected by Cornelia de Lange Syndrome (CdLS). In an effort to increase the amount of knowledge we have about children affected by CdLS and develop guidelines for educators and families, an online survey asking parents about their experiences in the education system and coordination with the medical system will establish a baseline of knowledge.

CdLS affects approximately 1:10,000 live births and is a congenital syndrome, meaning it is present from birth. Most of the signs and symptoms may be recognized at birth or shortly thereafter. A child need not demonstrate each and every sign or symptom for the diagnosis to be made.

As with other syndromes, individuals with CdLS strongly resemble one another. Common characteristics include: low birthweight (often under five pounds), slow growth and small stature, and small head size (microcephaly). Typical facial features include thin eyebrows which frequently meet at midline (synophrys), long eyelashes, short upturned nose and thin, downturned lips. Other frequent findings include excessive body hair (hirsutism), small hands and feet, partial joining of the second and third toes, incurved fifth fingers, gastroesophageal reflux, seizures, heart defects, cleft palate, bowel abnormalities, feeding difficulties, and developmental delay. Limb differences, including missing limbs or portions of limbs, usually fingers, hands or forearms, are also found in some individuals.

Mental retardation is usually present and ranges from mild to profound. The majority of cases fall in the mild to moderate range. Language ability is often affected, and many children do not communicate verbally at all.

This population has different educational and medical needs. For this reason we are interested in the range of services they are receiving, and if these services are meeting their needs. The data collected will inform professionals in the field and provide parents with an understanding of what others are experiencing. In addition, the information gathered will inform parents and other providers about this unique population about whom little is currently known.

## **METHODOLOGY**

**Procedure:** The CdLS-USA Foundation is the only resource for parents nationwide that provides support and information for families affected by CdLS (<http://www.cdlsusa.org/>). Their continued commitment to this population triggered this research project. The CdLS foundation has many involved board members who helped in the formation of the survey. In addition, the foundation distributed the survey link to their population in the form of email, through their newsletter, and by posting it on their web site.

In June 2007 a recruitment email/letter was emailed to approximately 590 families. The recruitment letter described the purpose of the study, the types of questions that would be asked and invited parents to click on a link to complete the survey online, or request a paper copy. One month later a reminder email to complete the survey was sent to these families. In August 2007 a newsletter was distributed to 2,000 families containing a recruitment flyer for the study. In September 2007 a final reminder email was sent out to the 590 families indicating that it was their last chance to participate, and the survey would be closed in October. Following study protocol, the online survey was closed on October 15, 2007.

**Data Collection:** The survey was completed online through Survey Monkey. No paper copies were requested. Participation was voluntary, and respondents had the option of bypassing any questions they did not want to answer. A total of 224 surveys were completed before the survey was closed to further data collection on October 15, 2007. Parents of a child age birth to 21 with CdLS were eligible to participate in the study.

**Questionnaire:** The questionnaire was developed in collaboration with the CdLS foundation and their advisory board as well as the University of Connecticut Center on Disabilities (see Appendix A for a copy of the survey). This survey was self-administered online and designed to gather information about the experiences of families with a child with CdLS and their service provision.

Questions were grouped according to 9 topic areas: (a) Background of Child; (b) Child Health and Medical Care; (c) Child Behavior and Development; (d) Child Learning Style; (e) Child Services; (f) Child Educational Placement; (g) Quality of Life; (h) Additional Information; and (i) Background Information.

**Data Management:** Surveys were completed by respondents online. Data was exported from the survey website to Excel and then SPSS.

**Data Analysis:** Descriptive statistics (means, frequencies, and percentages) were calculated for the quantitative data. Qualitative answers were coded and themed. The results from the data analysis of the survey are presented in the following results section.

## **RESULTS**

The results from the survey of 224 parents with children with CdLS is presented in the following section in order of survey topics: (a) Background of Child; (b) Child Health and

Medical Care; (c) Child Behavior and Development; (d) Child Learning Style; (e) Child Services; (f) Child Educational Placement; (g) Quality of Life; and (h) Background Information.

### **A: Background of Child**

The mean age of the parent completing the survey was 40 years with a standard deviation of 9.15, ranging from 19 to 66 years of age. The parent completing the survey was most commonly the mother (88%), followed by the father (7%), while the “other” group was comprised of adoptive parents, foster parents, and grandparents (5%).

The average age of the child was 9 years with a standard deviation of 5.68, ranging from 2 months to 20 years of age. Just over half of the children were female (61%), while 39% were male. Almost half of the children were diagnosed with CdLS at birth (42%), with 20% diagnosed in the first 6 months, 10% diagnosed when they were 7-12 months, 14% when they were 1-2 years old, 7% when they were 2-3 years old and 9% when they were over 3 years. The diagnosis was most commonly made by a geneticist (76%), followed by a pediatrician (13%), a neonatologist or neurologist (4%), “other”, which included the CdLS foundation (3%), an obstetrician (2%), and NICU Doctors (2%).

Seventy-two percent or 161 parents suspected that there was something wrong before the diagnosis. Within this group, most believed something was wrong because of their child’s low birth weight (62%), small stature (58%), slow growth (57%), poor feeding (52%), small head size (52%) and other various reasons listed in Table 1.

Table 1. Conditions that caused parent to suspect something was wrong with child (n=161)

<b>Condition</b>	<b>Frequency</b>	<b>Percent</b>
Low birth weight	100	62%
Small stature	94	58%
Slow growth	92	57%
Poor feeding	84	52%
Small head size	84	52%
Excessive body hair	78	48%
Eyebrows	73	45%
Small hands/feet	67	42%
Long eye lashes	57	35%
Low pitched crying	57	35%
Low set ears	48	30%
Curved fingers	46	29%
Strange different behavior	46	29%
Upturned nose	45	28%
Down turned lips	38	24%
Partial joining of toes	30	19%
Developmental delays	30	19%
Visual gaze averting behaviors	26	16%
Sensitive to touch	25	16%
Lack of feeling pain	22	14%
Missing fingers	6	4%
Other (Ex: Reflux, limb deformities, prenatal issues)	59	37%

## **B: Child Health and Medical Care**

Parents were asked about current health conditions their child experiences. Parents most commonly noted that their child experiences Gastroesophageal reflux disease (63%), feeding problems (43%), hearing loss (42%), and dental problems (41%) among other health conditions presented in Table 2.

Table 2. Current Health Conditions of Child. (n=224)

<b>Condition</b>	<b>Frequency</b>	<b>Percent</b>
Gastroesophageal reflux disease	140	63%
Feeding problems	96	43%
Hearing loss	93	42%
Dental problems	91	41%
Physical deformities	74	33%
Ear infections	69	31%
Heart problems/defect	65	29%
Vision problems: myopia (near sighted)	61	27%
Prescribed hearing aid	59	26%
Tube Fed	58	26%
Vision problems: Prescribed glasses	54	24%
Vision problems: general vision	47	21%
Wears hearing aid	42	19%
Vision problems: Wears glasses	36	16%
Vision problems: Blepharitis	27	12%
Cleft palate	25	11%
Seizures	24	11%
Vision problems: Light sensitive	16	7%
Vision problems: hyperopia (far sighted)	14	6%
Vision problems: Cortical/cerebral visual impairment	7	3%
Other (ex: behavioral, developmental, bladder)	85	38%

Most parents indicated that there is a place they usually contact when their child is sick (81%), 11% noted that there is more than one place, 6% indicated that there is no such place, and 1% did not know. The most common place to contact when their child is sick is their pediatrician (81%), followed by the hospital (14%), Herbalist (2%), GI doctor (2%) and nurse (1%). Almost all parents noted that there is a personal doctor or nurse that is a health provider for their child and knows them the best (91%), while a few parents did not have such a doctor (8%), and one person noted that they did not know. Most parents felt that it is not a problem to get a referral to a specialist that their child needed to see (66%), while 11% of children did not need to see a specialist in the last 12 months, and 8% of parents noted that they do not need referrals. A small group of parents noted difficulty in receiving referrals for their child: 14

parents or 6% noted that it was a big problem, and 17 parents or 8% noted that it was a small problem.

A series of questions asked about care coordination. Almost half of parents (45%) noted that in the past 12 months there was a time that their child needed care coordination among different health care providers and services. Of those who stated that care coordination was needed, 64% indicated that all the necessary coordination was received, and 28% reported that they did not receive all the coordination that was needed. Of those parents who had a care coordinator, most were reported to be a case manager or coordinator (34%), while others were a doctor (31%), the parent themselves (16%), a nurse (8%), a social worker (6%) or an early interventionist (3%). Regarding the frequency of this service, parents reported that someone helps coordinate their child's care among his/her different providers and services never (46%), sometimes (38%), usually (5%), and always (8%), while 2% indicated they did not know.

Overall, parents indicated that they were not especially satisfied with the help they have received in coordinating their child's care: 32% were very satisfied, 27% somewhat satisfied, 10% somewhat dissatisfied, 10% very dissatisfied, and 21% did not know how satisfied they were.

When asked how well they think their child's doctors and other health care providers communicate with each other about their child's care, answers were spread across the board: excellent (16%), very good (20%), good (23%), fair (19%), poor (12%), communication not needed (9%), and did not know (3%). When asked how well they think their child's doctors communicate with their school, early intervention program, child care providers or vocational program, parents indicated that communication is: excellent (11%), very good (10%), good (13%), fair (21%), poor (20%), not needed (21%), and did not know (5%).

### C: Child Behavior and Development

Parents were asked to describe their child’s current behavior and development. Table 3 describes the child behavior, how often the child engages in the behavior and the average age at which the child did this behavior frequently.

Table 3. Child Behavior and Developmental Tasks in Average Months.

	Does Not Do At All	Does Every Once in a While	Does Some times	Does A Lot	At what age did your child first do this a lot? Mean (S.D.)
Sitting or crawling	20 13%	7 5%	10 7%	114 16%	16 months (15)
Walking	55 32%	5 3%	9 5%	103 60%	25 months (19)
Going up stairs	57 33%	16 9%	21 12%	78 45%	28 months (29)
Running	72 42%	11 6%	24 14%	66 38%	31 months (27)
Feeds self with spoon/fork	58 33%	19 11%	25 14%	76 43%	29 months (29)
Drinks from cup by self	58 33%	7 4%	13 8%	96 55%	25 months (27)
Takes clothes off at appropriate times (e.g., going to bed)	84 49%	21 12%	22 13%	46 27%	41 months (41)
Puts clothes on by self	98 57%	18 11%	20 12%	36 21%	48 months (45)
Communicates using objects, gestures, pictures, icons, communication device	43 26%	30 18%	34 21%	58 35%	23 months (22)
Communicates using eye gaze	38 25%	31 20%	37 24%	46 30%	23 months (27)
Communicates using sign language	73 46%	35 22%	31 19%	21 13%	27 months (21)
Communicates using distinctive sounds	38 25%	20 13%	50 33%	46 30%	22 months (24)
Communicates using words	91 51%	13 7%	17 10%	57 32%	34 months (27)

	Does Not Do At All	Does Every Once in a While	Does Some times	Does A Lot	At what age did your child first do this a lot? Mean (S.D.)
Communicates using complete sentences (more than 3 words)	110 63%	6 3%	16 9%	43 25%	53 months (38)
My child responds to his/her name	19 10%	18 10%	35 19%	115 62%	18 months (22)
My child understands yes/no	29 16%	13 7%	51 28%	88 49%	19 months (22)
Recite alphabet	120 69%	8 5%	20 12%	25 15%	50 months (37)
Write first name	118 70%	4 2%	12 7%	36 21%	50 months (43)
Write full name	130 76%	9 5%	7 4%	26 15%	
If another child has a toy my child wants, my child takes the toy	61 35%	47 27%	50 28%	19 11%	
If another child has toy my child wants, my child requests toy without taking	94 55%	28 16%	40 23%	10 6%	
Is aggressive or hurts others	104 57%	39 21%	30 17%	9 5%	
Gets along with others	16 9%	10 6%	42 23%	114 63%	
Hurts himself/herself	72 39%	43 23%	47 25%	24 13%	
My child makes friends on his/her own (not through arranged play dates by parents)	86 49%	39 22%	27 15%	23 13%	
My child has a friend (may call child's house or friend calls my child)	117 69%	18 11%	20 12%	15 9%	
My child has had and/or gone to a sleepover at the home of classmate	156 89%	11 6%	5 3%	3 2%	
Moody or irritable	27 15%	55 30%	64 35%	38 21%	
Quiet or shy	48 26%	48 26%	71 38%	19 10%	

## D: Child Learning Style

Parents were asked to think about their child’s learning style when he or she is discovering new things. A series of eight questions were asked regarding the child’s learning style, and the results are presented in Table 4.

Table 4. Child Learning Style.

	Not At All True	A Little True	Somewhat True	Very True
When visiting a new place, my child independently explores it on his/her own.	42 23%	32 17%	47 25%	66 35%
When visiting a new place, my child remains near me or someone familiar.	45 24%	52 28%	42 23%	46 25%
My child enjoys learning new things. For example, learning the words to a new song, riding a bike, a new toy.	37 20%	43 23%	44 24%	60 33%
My child requests assistance when encountering new things, such as a toy, asking a question about a new subject in class, etc.	57 32%	47 26%	44 24%	32 18%
My child likes receiving praise. For example, when teacher says “good job” or parent says “great work” when learning a new skill.	23 12%	24 13%	25 13%	115 62%
My child tells me about their day.	109 61%	33 18%	25 14%	13 7%
When my child is explaining something, and I do not understand, he/she gets frustrated.	74 44%	32 19%	29 17%	32 19%
My child is easily upset by simple demands, such as no, stop, etc.	85 48%	45 25%	28 16%	21 12%

## E: Child Services

Information about the types of services a child has never received, has received in the past and is currently receiving was gathered from parents. Table 5 displays these results. The most commonly provided service that a child currently receives is special education (84%), followed by speech and language therapy (78%), occupational therapy (65%) and physical

therapy (63%), among many other services listed in Table 5. The most commonly provided services in the past were early intervention (70%), feeding therapy (44%), physical therapy (33%) and occupational therapy (29%), among other services presented in Table 5. The services least frequently provided are hippotherapy (90%), cranial-sacral therapy (90%), Community assistance program/MR (87%), and behavior therapy (82%), followed by many other services listed in Table 5.

Table 5. Types of Services Child Never, Recently, or Currently Received

<b>Service</b>	<b>Has Never Received</b>	<b>Received in the Past</b>	<b>Currently Receives</b>
Respite care	111 56%	30 16%	51 27%
Physical therapy	9 5%	64 33%	123 63%
Speech and language therapy	11 6%	32 17%	150 78%
Special education	21 11%	10 5%	157 84%
Early intervention	11 6%	135 70%	46 24%
Feeding therapy	79 42%	82 44%	27 14%
Child care/respite care	96 50%	44 23%	51 27%
Occupational therapy	11 6%	56 29%	126 65%
Mental health services	148 78%	17 9%	26 14%
Nursing care	130 68%	39 21%	21 11%
Nutritional therapy	117 62%	55 29%	16 9%
Behavior therapy	158 82%	18 9%	17 9%
Adaptive equipment/prosthesis	112 58%	33 17%	48 25%
CAP/MR (Community Assistance Program)	165 87%	5 3%	20 11%
Medicaid	62 32%	16 8%	114 59%

Service	Has Never Received	Received in the Past	Currently Receives
Computer Assisted Communication	136 72%	35 19%	18 10%
Cranial-sacral therapy	170 90%	16 9%	3 2%
Music therapy	130 67%	32 17%	32 17%
Hippotherapy	170 90%	10 5%	9 5%
“Other” (10 parents indicated some “other” examples: swimming lessons, sensory integration, massage therapy)			

### F: Child Educational Placement

Based on the age of their child, parents completed a section on educational placement where they could choose from Early Intervention (birth to three years), Preschool (3 to 5 years), Elementary school (grades K through 5) or Middle and High school.

*Early Intervention.* There were 95 parents who had children in Early Intervention (birth to three years), and these findings are presented in Table 6. Parents indicated that most IFSPs include outcomes that are important to them (76%), that EI staff build on their child’s strengths (72%), and that staff are provided as listed on their IFSP (80%). However numbers are a bit lower regarding EI staff helping them work with their child (38%), indicating that family-centered practices are not being used. In addition, only 13% of parents noted that staff work closely with medical professionals who are treating their child, indicating low coordination of care.

Only 15% of staff were knowledgeable about CdLS. Parents were asked how staff became knowledgeable, and the most common ways for staff to get information about CdLS were the CdLS foundation/web site (32, 52%), the parent themselves (19, 31%), having previous experience with another child (4, 7%), and their doctors (5, 8%), while one person noted that the

therapist working with their child received training from their educational and school background on CdLS.

Table 6. Children in Early Intervention (Birth to Three years) (n=95).

<b>Early Intervention (Birth to 3 Years)</b>	<b>Not At All True</b>	<b>A Little True</b>	<b>Somewhat True</b>	<b>Very True</b>
My child's Individualized Family Service Plan (IFSP) includes outcomes that are important to me	5 5%	5 5%	13 14%	72 76%
Early Intervention staff recognize/build on my child's strengths	3 3%	5 6%	17 19%	65 72%
Early Intervention staff are provided as listed on my child's Individualized Family Service Plan (IFSP)	3 3%	2 2%	13 14%	72 80%
Early Intervention staff are knowledgeable about Cornelia de Lange Syndrome	24 26%	35 38%	20 22%	14 15%
Early Intervention staff help me learn how to work with my child	9 10%	16 17%	33 35%	36 38%
School staff work closely with medical professionals treating my child	41 45%	25 27%	14 15%	12 13%

*Preschool.* There were 63 parents who had a child in preschool (3 to 5 years), and these findings are presented in Table 7. Most of the students spend the majority of their time in a separate classroom (43, 68%), and 20 students spend most of their time in a general education or regular education classroom (32%). Parents noted that most preschool staff communicate regularly with them (76%), that their IEPs include goals that are important to them (72%), and that their child's teacher/therapist encourages their child to do their best (79%). Parents did report that only 21% of school staff work closely with medical professionals who are treating their child, indicating low coordination of care.

Only 13% of staff were knowledgeable about CdLS. Parents were asked how staff became knowledgeable, and the most common ways for staff to get information about CdLS

were the CdLS foundation/web site (20, 49%), the parent themselves (16, 39%), having previous experience with another child (3, 7%), and their doctors (2, 5%).

Table 7. Child Educational Placement Preschool (3 to 5 years ) (n=63)

<b>Preschool (3 to 5 Years)</b>	Not At All True	A Little True	Somewhat True	Very True
Preschool staff communicate regularly with me	2 3%	3 5%	9 15%	45 76%
Preschool staff are knowledgeable about Cornelia de Lange Syndrome	15 25%	19 32%	18 30%	8 13%
My child's Individualized Education Plan (IEP) includes goals that are important to me	2 3%	3 5%	12 20%	43 72%
My child's teacher/therapists encourage my child to do his/her best	2 3%	0 0%	11 18%	48 79%
School staff work closely with medical professionals treating my child	23 38%	17 28%	8 13%	13 21%

*Elementary School.* There were 76 parents who had a child in elementary school (grades K to 5), and these findings are presented in Table 8. Most of the students spend the majority of their time in a separate classroom (55, 74%), and 19 students spent most of their time in a general education or regular education classroom (26%). Parents are reported that only 29% of children have classmates that are his or her friends, indicating a great need for social interactions and friendship making. School facilities are fully accessible to most children (71%), and IEP goals are important to parents (71%). Parents did report that only 14% of school staff work closely with medial professionals who are treating their child, indicating low coordination of care.

Only 18% of staff were knowledgeable about CdLS. Parents were asked how staff became knowledgeable, and the most common ways for staff to get information about CdLS were the parent themselves (29, 50%), the CdLS foundation/web site (25, 43%), and having

previous experience with another child (2, 3%), while one person mentioned a conference or training that the teacher attended, and one person noted the Kennedy Krieger foundation.

Table 8. Child Educational Placement Elementary School (Grades K to 5) (n=76)

<b>Elementary School (Grades K to 5)</b>	<b>Not At All True</b>	<b>A Little True</b>	<b>Somewhat True</b>	<b>Very True</b>
My child has classmates that are his/her friends	8 11%	21 28%	24 32%	22 29%
School facilities are fully accessible to my child	3 4%	2 3%	17 22%	54 71%
My child's Individualized Education Plan (IEP) includes goals that are important to me	1 1%	2 3%	19 25%	53 71%
My child's teacher/therapists are knowledgeable about Cornelia de Lange Syndrome	13 17%	29 38%	20 26%	14 18%
School staff work closely with medical professionals treating my child	31 43%	19 26%	13 18%	10 14%

*Middle School and High School.* There were 64 parents who had a child in middle or high school, and these findings are presented in Table 9. Most of the students spend the majority of their time in a separate classroom (50, 86%), and 8 students spend most of their time in a general education or regular education classroom (14%). Parents were asked if their child's IEP included everything it should for teaching reading and writing skills. 40% felt this to be very true, but 26% felt it was not at all true. Thirty-five percent of parents felt that their child would graduate from high school with their classmates, while 35% felt this was not true at all. Almost half of parents (47%) felt that school staff do not work closely with medical professionals who are working with their child. Most parents do not receive support or services from vocational rehab counselors (78%), and only 24% receive support form other community supports or programs.

Only 13% of staff were knowledgeable about CdLS. Parents were asked how staff became knowledgeable, and the most common ways for staff to get information about CdLS were the parent themselves (27, 63%), the CdLS foundation/web site (9, 21%), and staff training or attendance at the annual conference (3, 7%). In addition, two parents mentioned doctors involved in training or helping staff become knowledgeable, one parent noted the school had experience with other children with CdLS, and one parent mentioned the Kennedy Krieger foundation.

Table 9. Child Educational Placement Middle and High School (n=64)

<b>Middle and High School</b>	Not At All True	A Little True	Somewhat True	Very True
My child's teachers/therapists are knowledgeable about Cornelia de Lange Syndrome	13 20%	17 27%	26 41%	8 13%
My child's Individualized Education Plan (IEP) includes everything it should for teaching reading and writing skills	15 26%	10 17%	10 17%	23 40%
I expect my child to graduate from high school with his/her classmates	21 35%	11 18%	7 12%	21 35%
School staff work closely with medical professionals treating my child	28 47%	22 37%	6 10%	4 7%
I receive support and/or services from Vocational Rehab counselors	47 78%	6 10%	5 8%	2 3%
I receive support and/or services from other agencies and community programs	24 39%	11 18%	12 19%	15 24%

## **G: Quality of Life**

Parents were asked about their own and their family's quality of life. Questions and responses are shown in Table 10. Parents responded that they are happy with how their life is going (44%), their family likes spending time together (83%), they enjoy relaxing with family members (73%), and that their family has the best life possible (34%). Almost half reported that family meal times are not stressful (44%), several parents stated that it is hard for them to get

going (10%), very few parents noted that family members drive each other crazy (2%) and only a small number of parents let little things bother them (4%).

Table 10. Quality of Life.

	Not At All True	A Little True	Somewhat True	Very True
I am happy with how my life is going	4 2%	15 18%	86 46%	83 44%
Family meal times are stressful	82 44%	45 24%	43 23%	17 9%
Our family likes spending time together	1 .5%	9 5%	21 11%	156 83%
Some days it is hard for me to “get going”	54 29%	62 33%	52 28%	19 10%
I enjoy relaxing with other family members	4 2%	9 5%	37 20%	137 73%
Family members drive each other crazy	70 38%	71 38%	41 22%	3 2%
I let little things bother me	71 38%	85 46%	23 12%	8 4%
Our family has the best life possible	16 9%	18 10%	90 48%	63 34%

## H: Background Information

Families resided in a variety of locations in the United States. Most lived in an urban area (40%), followed by a city (34%) and a rural area (26%). The majority of parents were white (75%), 9% were Hispanic, 5% were African American, 1% were Asian, and several were Native American. Most parents stated that they were non-denominational/other or multiple religions (36%), followed by Catholic (24%), Christian (21%), Baptist (10%), and a few parents wrote none or not applicable (9%). The majority of parents were married (81%), followed by divorced or separated (9%), never married (6%), in a co-parenting relationship (3%), and one who was a widow. Parent responses were evenly distributed between not working (38%), working full time

(38%) and working part time (24%). Most spouses were working full time (76%), a few were not working (9%), and 4% were working part time.

Parents were also asked about their highest educational attainment. One parent had less than a high school education, 11% had a high school diploma, 36% had some college, 40% had a college degree, 10% had a masters degree, and 2% had a PhD, MD or JD. Income ranged from less than \$19,000 to more than \$150,000, with most family incomes falling between \$30,000 and \$39,000. Parents represented 39 different states with the highest response from Texas (10%).

## **APPENDIX A: SURVEY**

## Parent Survey of Children with Cornelia de Lange Syndrome

Thank you for taking the time to complete this survey on your family member with Cornelia de Lange Syndrome. The information will help us better understand the health and development of children with this condition.

---

### Section A: Please Tell Us About Your Child

1. Your age in years: \_\_\_\_\_
2. Relationship to child:
  - Mother
  - Father
  - Other (please describe) \_\_\_\_\_
3. What is your child's current age? \_\_\_\_\_
4. Is your child:  Male  Female
5. At what age was your child diagnosed with Cornelia de Lange Syndrome?
  - Prenatally
  - At Birth
  - 1-6 Months
  - 7-12 Months
  - 1-2 Years
  - 2-3 Years
  - 3+ Years
6. Who made the diagnosis?
  - Obstetrician
  - Geneticist
  - Pediatrician
  - Other (please describe \_\_\_\_\_)
7. Did you suspect something was wrong before the diagnosis?
  - Yes
  - No

7a. If yes, please check any of the following things that made you suspect something might be wrong:

  - Low birth weight

- Upturned nose
- Curved fingers
- Long eye lashes
- Low set ears
- Low pitched crying
- Small stature
- Down turned lips
- Excessive body hair
- Partial joining of toes
- Poor feeding
- Lack of feeling of pain
- Small head size
- Small hands/feet
- Eyebrows
- Slow growth
- Strange/different behavior
- Sensitive to touch
- Visual gaze averting behaviors
- Other (please describe)\_\_\_\_\_

## Section B: Child Health and Medical Care

1. Please check any of the following conditions that your child currently experiences.
  - Feeding problems
  - Seizures
  - Dental problems
  - Gastroesophageal reflux disease
  - Tube fed
  - Vision problems: General vision problems
  - Vision problems: Myopia (near-sighted)
  - Vision problems: Hyperopia (far-sighted)
  - Vision problems: Cortical/cerebral visual impairment
  - Vision problems: Blepharitis
  - Vision problems: Light sensitive
  - Vision problems: Prescribed glasses
  - Vision problems: Wears glasses
  - Physical deformities
  - Ear infections
  - Heart problems/defect
  - Hearing loss
  - Prescribed hearing aid
  - Wears hearing aid
  - Cleft palate
  - Other (please describe) \_\_\_\_\_
  
2. Is there a place you usually contact when your child is sick?
  - Yes
  - There is no place
  - There is more than one place
  - Don't knowIf there is a place(s), please list: \_\_\_\_\_
  
3. A personal doctor or nurse is the health provider who knows your child the best. Do you have ONE person that you think of as your child's personal doctor or nurse?
  - Yes
  - No
  - Don't know
  
4. In the past 12 months, how much of a problem, if any, was it to get a referral to a specialist that your child needed to see?
  - A big problem
  - A small problem
  - Not a problem
  - Child did not need to see a specialist in the past 12 months
  - Don't need referrals
  - Don't know

A case manager or a case coordinator is a person who assists in coordinating care to make sure that your child gets the services that he/she needs.

5. During the past 12 months, was there any time when your child needed care coordination among different health care providers and services that your child uses?

- Yes
- No
- Don't know

5a. If yes, Did you or your family receive all the professional care coordination that was needed?

- Yes
- No
- Don't know

5b. If you have a care coordinator, what is their title/position? \_\_\_\_\_

6. How often does a person help you coordinate your child's care among his/her different providers and services? Would you say:

- Never
- Sometimes
- Usually
- Always
- Don't know

7. Overall how satisfied are you with the help you have received in coordinating child's care?

- Very satisfied
- Somewhat satisfied
- Somewhat dissatisfied
- Very dissatisfied
- Don't know

8. How well do you think your child's doctors and other health care providers communicate with each other about your child's care? Would you say their communication is:

- Excellent
- Very Good
- Good
- Fair
- Poor
- Communication not needed
- Don't know

9. How well do think your child's doctors and other health care providers communicate with his/her school, early intervention program, child care providers, or vocational rehabilitation program? Would you say their communication is:
- Excellent
  - Very Good
  - Good
  - Fair
  - Poor
  - Communication not needed
  - Don't know

### Section C: Child Behavior and Development

Please indicate which response best describes your child's behavior and development at this time. Skip any item that does not apply to your child (e.g., because of his or her age).					
	Does Not Do At All	Does Every Once in a While	Does Some times	Does A Lot	At what age did your child first do this a lot?
a. Sitting or crawling	1	2	3	4	( ) months
b. Walking	1	2	3	4	( ) months
c. Going up stairs	1	2	3	4	( ) months
d. Running	1	2	3	4	( ) months
e. Feeds self with spoon/fork	1	2	3	4	( ) months
f. Drinks from cup by self	1	2	3	4	( ) months
g. Takes clothes off at appropriate times (e.g., going to bed)	1	2	3	4	( ) months
h. Puts clothes on by self	1	2	3	4	( ) months
i. Communicates using objects, gestures, pictures, icons, communication device	1	2	3	4	( ) months
j. Communicates using eye gaze	1	2	3	4	( ) months
k. Communicates using sign language	1	2	3	4	( ) months
l. Communicates using distinctive sounds	1	2	3	4	( ) months
m. Communicates using words	1	2	3	4	( ) months
n. Communicates using complete sentences (more than 3 words)	1	2	3	4	( ) months
o. My child responds to his/her name	1	2	3	4	( ) months
p. My child understands yes/no	1	2	3	4	( ) months

	Does Not Do At All	Does Every Once in a While	Does Some times	Does A Lot	At what age did your child first do this a lot?
q. Recite alphabet	1	2	3	4	( ) months
r. Write first name	1	2	3	4	( ) months
s. Write full name	1	2	3	4	( ) months
t. If another child has a toy my child wants, my child takes the toy	1	2	3	4	
u. If another child has toy my child wants, my child requests toy without taking	1	2	3	4	
v. Is aggressive or hurts others	1	2	3	4	
w. Gets along with others	1	2	3	4	
x Hurts himself/herself	1	2	3	4	
y. My child makes friends on his/her own (not through arranged play dates by parents)	1	2	3	4	
z. My child has a friend (may call child's house or friend calls my child)	1	2	3	4	
aa. My child has had and/or gone to a sleepover at the home of classmate	1	2	3	4	
bb. Moody or irritable	1	2	3	4	
cc. Quiet or shy	1	2	3	4	

By “friends” we mean that the children like each other and play together.

1. How many friends did you invite to your child’s last birthday party?

\_\_\_\_\_ total # of friends  
\_\_\_\_\_ # of friends with disabilities  
\_\_\_\_\_ # of friends without disabilities

2. How many friends attended your child’s last birthday party?

\_\_\_\_\_ total # of friends attended  
\_\_\_\_\_ # of friends with disabilities  
\_\_\_\_\_ # of friends without disabilities

3. How many birthday parties was your child invited to last year?

\_\_\_\_\_ # of parties invited to

4. How many birthday parties did your child attend last year?

\_\_\_\_\_ # of parties attended

### Section D: Child Learning Style

This section of the survey asks you to think about your child’s learning style while doing the things he or she is learning to do at this time. Please indicate the extent to which each statement is true.				
	Not At All True	A Little True	Somewhat True	Very True
a. When visiting a new place, my child independently explores it on his/her own	1	2	3	4
b. When visiting a new place, my child remains near me or someone familiar	1	2	3	4
c. My child enjoys learning new things. For example, learning the words to a new song, riding a bike, a new toy.	1	2	3	4
d. My child requests assistance when encountering new things, such as a toy, asking a question about a new subject in class, etc.	1	2	3	4
e. My child likes receiving praise. For example, when teacher says “good job” or parent says “great work” when learning a new skill.	1	2	3	4
f. My child tells me about their day	1	2	3	4
g. When my child is explaining something, and I do not understand, he/she gets frustrated.	1	2	3	4
h. My child is easily upset by simple demands, such as no, stop, etc.	1	2	3	4

### Section E: Child Services

Please indicate whether or not your child has in the past or currently receives any of the following services.			
Services	Has Never Received	Received in the Past	Currently Receives
a. Respite care	1	2	3
b. Physical therapy	1	2	3
c. Speech and language therapy	1	2	3
d. Special education	1	2	3
e. Early intervention	1	2	3
f. Feeding therapy	1	2	3
g. Child care/respite care	1	2	3
h. Occupational therapy	1	2	3
i. Mental health services	1	2	3
j. Nursing care	1	2	3
k. Nutritional therapy	1	2	3
l. Behavior therapy	1	2	3
m. Adaptive equipment/prosthesis	1	2	3
n. CAP/MR (Community Assistance Program)	1	2	3
o. Medicaid	1	2	3
p. Computer Assisted Communication	1	2	3
q. Cranial-sacral therapy	1	2	3
r. Music therapy	1	2	3
s. Hippotherapy	1	2	3
t. Other (please describe) _____	1	2	3

**Section F: Child Educational Placement**

Please complete the questions under the section that best describes your child’s current early intervention or educational placement.

<b>1. Early Intervention (Birth to 3 Years)</b>	Not At All True	A Little True	Somewhat True	Very True
a. My child’s Individualized Family Service Plan (IFSP) includes outcomes that are important to me	1	2	3	4
b. Early Intervention staff recognize/build on my child’s strengths	1	2	3	4
c. Early Intervention staff are provided as listed on my child’s Individualized Family Service Plan (IFSP)	1	2	3	4
d. Early Intervention staff are knowledgeable about Cornelia de Lange Syndrome	1	2	3	4
e. Early Intervention staff help me learn how to work with my child	1	2	3	4
f. School staff work closely with medical professionals treating my child	1	2	3	4
g. If staff are knowledgeable about CdLS and/or have taught you to work with your child, how did they obtain this information?				
_____				
_____				
_____				

<b>2. Preschool (3 to 5 Years)</b>	Not At All True	A Little True	Somewhat True	Very True
a. Where does your child spend most of his/her time? Circle one. General Education/regular classroom Separate classroom setting				
b. Preschool staff communicate regularly with me	1	2	3	4
c. Preschool staff are knowledgeable about Cornelia de Lange Syndrome	1	2	3	4
d. My child’s Individualized Education Plan (IEP) includes goals that are important to me	1	2	3	4
e. My child’s teacher/therapists encourage my	1	2	3	4

child to do his/her best				
f. School staff work closely with medical professionals treating my child	1	2	3	4
g. If staff are knowledgeable about CdLS and/or have taught you to work with your child, how did they obtain this information?				
_____				
_____				
_____				

<b>3. Elementary School (Grades K to 5)</b>	Not At All True	A Little True	Somewhat True	Very True
a. Where does your child spend most of his/her time? Circle one. General Education/regular classroom Separate classroom setting				
b. My child has classmates that are his/her friends	1	2	3	4
c. School facilities are fully accessible to my child	1	2	3	4
d. My child's Individualized Education Plan (IEP) includes goals that are important to me	1	2	3	4
e. My child's teacher/therapists are knowledgeable about Cornelia de Lange Syndrome	1	2	3	4
f. School staff work closely with medical professionals treating my child	1	2	3	4
g. If staff are knowledgeable about CdLS and/or have taught you to work with your child, how did they obtain this information?				
_____				
_____				
_____				

<b>4. Middle and High School</b>	Not At All True	A Little True	Somewhat True	Very True
a. Where does your child spend most of his/her time? Circle one. General Education/regular classroom Separate classroom setting	1	2	3	4
b. My child's teachers/therapists are knowledgeable about Cornelia de Lange Syndrome	1	2	3	4
c. My child's Individualized Education Plan (IEP) includes everything it should for teaching reading and writing skills	1	2	3	4
d. I expect my child to graduate from high school with his/her classmates	1	2	3	4
e. School staff work closely with medical professionals treating my child	1	2	3	4
f. I receive support and/or services from Vocational Rehab counselors	1	2	3	4
g. I receive support and/or services from other agencies and community programs	1	2	3	4
<p>If staff are knowledgeable about CdLS and/or have taught you to work with your child, how did they obtain this information?</p> <hr/> <hr/> <hr/>				



## Section I: Background Information

Please tell us about yourself and your family by completing the following.

1. We live in a:
  - City
  - Urban area
  - Rural area
  
2. What is your race/ethnicity? (You may check all that apply)
  - White
  - Latino/Hispanic
  - Black or African American
  - Asian
  - Other: \_\_\_\_\_
  
3. Your religious affiliation(s): \_\_\_\_\_
  
4. Your current marital status:
  - married
  - never married
  - co-parenting relationship
  - divorced/separated
  - widowed/spouse passed away
  
5. Are you currently employed?
  - Yes-full-time
  - Yes-part-time
  - No
  
6. Is your spouse/partner currently employed?
  - Yes-full-time
  - Yes-part-time
  - No
  - N/A
  
7. What is your highest educational attainment?
  - Less than high school
  - High school
  - Some college
  - College (Degree in \_\_\_\_\_)
  - Master's (Degree in \_\_\_\_\_)
  - Doctorate/Ph.D./Ed.D./M.D./J.D. (Degree in \_\_\_\_\_)

8. In 2006, what was your approximate combined gross household income in U.S. dollars?

- |   |  |  |
|---|--|--|
| <input type="checkbox"/> Less than \$19,999 | <input type="checkbox"/> \$60,000-\$69,999   | <input type="checkbox"/> \$110,000-\$119,999 |
| <input type="checkbox"/> \$20,000-\$29,999  | <input type="checkbox"/> \$70,000-\$79,999   | <input type="checkbox"/> \$120,000-\$129,999 |
| <input type="checkbox"/> \$30,000-\$39,999  | <input type="checkbox"/> \$80,000-\$89,999   | <input type="checkbox"/> \$130,000-\$139,999 |
| <input type="checkbox"/> \$40,000-\$49,999  | <input type="checkbox"/> \$90,000-\$99,999   | <input type="checkbox"/> \$140,000-\$149,999 |
| <input type="checkbox"/> \$50,000-\$59,999  | <input type="checkbox"/> \$100,000-\$109,999 | <input type="checkbox"/> More than \$150,000 |

9. What state/territory do you currently live in?

<input type="checkbox"/> Alabama	<input type="checkbox"/> Illinois	<input type="checkbox"/> Nebraska	<input type="checkbox"/> South Carolina
<input type="checkbox"/> Alaska	<input type="checkbox"/> Indiana	<input type="checkbox"/> Nevada	<input type="checkbox"/> South Dakota
<input type="checkbox"/> Arizona	<input type="checkbox"/> Iowa	<input type="checkbox"/> New Hampshire	<input type="checkbox"/> Tennessee
<input type="checkbox"/> Arkansas	<input type="checkbox"/> Kansas	<input type="checkbox"/> New Jersey	<input type="checkbox"/> Texas
<input type="checkbox"/> California	<input type="checkbox"/> Kentucky	<input type="checkbox"/> New Mexico	<input type="checkbox"/> Utah
<input type="checkbox"/> Colorado	<input type="checkbox"/> Louisiana	<input type="checkbox"/> New York	<input type="checkbox"/> Vermont
<input type="checkbox"/> Connecticut	<input type="checkbox"/> Maine	<input type="checkbox"/> North Carolina	<input type="checkbox"/> Virgin Islands
<input type="checkbox"/> Delaware	<input type="checkbox"/> Maryland	<input type="checkbox"/> North Dakota	<input type="checkbox"/> Virginia
<input type="checkbox"/> District of Columbia	<input type="checkbox"/> Massachusetts	<input type="checkbox"/> Ohio	<input type="checkbox"/> Washington
<input type="checkbox"/> Florida	<input type="checkbox"/> Michigan	<input type="checkbox"/> Oklahoma	<input type="checkbox"/> West Virginia
<input type="checkbox"/> Georgia	<input type="checkbox"/> Minnesota	<input type="checkbox"/> Oregon	<input type="checkbox"/> Wisconsin
<input type="checkbox"/> Guam	<input type="checkbox"/> Mississippi	<input type="checkbox"/> Pennsylvania	<input type="checkbox"/> Wyoming
<input type="checkbox"/> Hawaii	<input type="checkbox"/> Missouri	<input type="checkbox"/> Puerto Rico	
<input type="checkbox"/> Idaho	<input type="checkbox"/> Montana	<input type="checkbox"/> Rhode Island	<input type="checkbox"/> Other: Specify_____

If you would like to participate in any of our follow up surveys or receive a copy of our results from this study please provide your contact information. We will also post our results on our

web site: <http://www.uconnucedd.org/index.htm>

Your contact information will not be connected to any of the information you have provided, it will be kept separate from your responses and will be kept confidential and locked and destroyed after 5 years of completion of this project.

- I would like to be contacted for other studies
- I would like to receive the results of this study

Name: \_\_\_\_\_

Address: \_\_\_\_\_

\_\_\_\_\_

Email: \_\_\_\_\_

Phone: \_\_\_\_\_