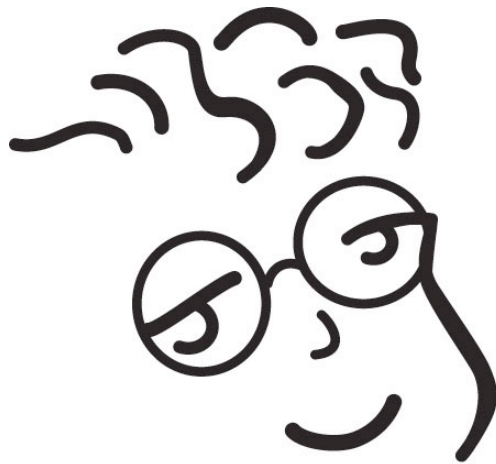


Cardio-Facio-Cutaneous Syndrome Registry



CFCinternational

**CFC International
183 Brown Road
Vestal, New York 13850
Phone: (607) 772-9666**

Web site: www.cfcsyndrome.org
Email: bconger@cfcsyndrome.org

You are being asked to complete a questionnaire about CFC Syndrome

Thank you for filling out this questionnaire. It is very important that you answer each question, even if your answer is “no” or “I do not know”. Please answer to the best of your ability. If you are not able to answer the questionnaire, try to ask a relative or friend to help. Questions can be e-mailed to: bconger@cfcsyndrome.org. As a last resort, please call CFC International at (607) 772-9666.

Purpose of the Questionnaire:

The purpose of this data collection is to learn more about the characteristics of the population affected by CFC Syndrome. The CFC syndrome includes a wide range of symptoms that we still need to learn about. The discovery of the CFC genes has given us an opportunity to understand how the genes and symptoms are related. Thank you for participating in this registry.

Confidentiality:

All information you provide to CFC International will remain confidential. The detailed information that you provide will allow CFC International medical advisors to understand the full spectrum of this condition that is essential to foster a strong research effort. If the information in the registry can support a specific research project, information will be shared without with only a BioBank ID number on the documents and all private identification removed. On occasion families benefit from understanding the experiences of other families who have addressed a specific medical or developmental concern. This allows us to match families. Families will be contacted to seek permission to share contact information.

Brenda Conger
President
CFC International, Inc.

Completed questionnaires (with photographs and copies of specialist reports) can be mailed to:

CFC International, Inc
183 Brown Road
Vestal, New York 13850
USA

Alternatively, the questionnaire can be completed electronically, which may be more convenient for overseas families. In MS Word, you can double-click wherever you need to fill in text. Please e-mail bconger@cfcsyndrome.org for an electronic copy, if you would prefer this option.

Volunteer Statement

- I understand that I can voluntarily enter and/or withdraw from this project without loss of benefits or treatment to which I might otherwise be entitled.
- I understand that CFC International, Inc. provides no institutional benefit or financial compensation, including payment of expenses associated with medical treatment.
- I understand that my child's name or photo will not be used in any publication resulting from this project without my consent.
- I understand that all records relative to this project will be treated in confidence, being available only to CFC International, Inc. and its authorized medical advisors and/or researchers.
- I understand that if I have any questions relating to this project that I can telephone Brenda Conger at (607) 772-9666 during evenings and on weekends.
- I have voluntarily agreed to participate in this research questionnaire. I understand the purpose of the study of CFC syndrome.

Print name of Patient or Parent/Legal Guardian:

Signature of Patient or Parent/Legal Guardian

Date

Photograph Release Form

Photographs of the CFC Syndrome adult or child would be helpful for comparisons and educational purposes. We request that you enclose at least three (3) photographs showing different ages of your child, that you would like to share with CFC International. Please sign this release form (indicating Yes or No as applicable) and send the photos along with this questionnaire. On the back of each photo, please write the name, birth date and age of your child in the photo at the time it was taken. Photos will not be returned.

I, _____
(Print name of person giving consent)

- Give consent for photographs to be used by CFC International, Inc for comparison and education YES / NO *
- Give consent for my child's photograph to be used on the CFC web site with only the first name YES / NO *

Number of photos included: _____

Signature

Date

* *Delete or circle "Yes" or "No" as applicable. If no deletions are made, it will be assumed your consent is being given for all 3 statements.*

Personal Contact details

CFC Individual's Name:

First

Middle

Last

Parent's Names:

Street or PO Box:

City, State, Zip:

Country:

Telephone:

E-Mail Address:

Siblings:

Name:

Date of Birth:

Contact Details for two people (family or friends) who would know how to reach you if you move:

Name:

Address:

City:

State/Zip:

Telephone:

Relationship:

Name:

Address:

City:

State/Zip:

Telephone:

Relationship:

Details of your Child's Physicians

Primary Care Physician

Name _____
Speciality _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

Geneticist

Name _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

Cardiologist

Name _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

Other Physicians *(eg Dermatologist; Endocrinologist; Neurologist; Ophthalmologist; ENT etc)*

Name _____
Speciality _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

Name _____
Speciality _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

Name _____
Speciality _____
Address _____
City, State, Zip _____ Country _____
Area Code & Telephone Number _____

CFC Syndrome Characteristics & Symptoms

Indicate the age at which any of the following characteristics were first experienced or noticed.
 If the characteristic is not apparent in your child, check the "N/A" column.
 Check the "Do not Know" column if you have no knowledge of the characteristic/symptom or are unsure whether your child is affected.

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st</i> <i>experienced</i> <i>or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
PRENATAL/PERINATAL					
POLYHYDRAMNIOS (too much amniotic fluid)					
GESTATIONAL DIABETES					
ULTRASOUNDS PERFORMED (any abnormalities noted)					
NUCHAL SKIN OR FOLD					
NEONATAL HYPOGLYCEMIA					
GESTATIONAL HYPERTENSION					
KNOWN EXPOSURES OR MATERNAL ILLNESS					
MOVEMENT IN UTERO (decreased or increased movement)					
GESTATION (weeks)					
MOTHER'S AGE AT CONCEPTION					
FATHER'S AGE AT CONCEPTION					
OTHER PRENATAL HISTORY					
HEART					
PULMONARY STENOSIS					
ASD (atrial septal defect)					
VSD (ventricular septal defect)					
HEART MURMUR					
HYPERTROPHIC CARDIOMYOPATHY					
ARRHYTHMIA, SLOW OR FAST (irregular heart beat)					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
IRREGULAR HEART BEAT WITH GENERAL ANESTHESIA					
OTHER HEART DEFECT (tricuspid valve, bicuspid valve, mitral valve)					
ECHOCARDIOGRAMS					
HEART SURGERY (please specify details in comments column)					
CRANIO FACIAL APPEARANCE					
DYSMORPHIC FEATURES					
BITEMPORAL NARROWING					
CHERUBISM (broad, thick lower jaw)					
DEPRESSED NASAL BRIDGE					
SHORT NOSE, BULBOUS TIP					
DOWNSLANTING PALPEBRAL FISSURES					
EPICANTHAL FOLD					
PTOSIS (droopy eye lids)					
LARGE HEAD					
FRONTAL BOSSING (high forehead)					
SHALLOW EYE SOCKET					
MOUTH (large or small)					
DENTAL CARIES (thin enamel or cavities)					
SIZE OF TONGUE (normal, large, small)					
HIGH ARCHED PALATE					
KERATOSIS PILARIS					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
HYPERKERATOSIS (thick skin)					
RED ITCHY SKIN					
DERMATITIS					
CAFÉ-AU-LAIT SPOTS					
NEVI (moles, birthmarks, etc)					
ICHTHYOSIS					
ECZEMA					
SUBCUTANEOUS LUMPS					
HEMANGIOMAS					
PAPILLOMA (small, warty lesions)					
CUTANEOUS LYMPHANGIOMA (wart like growth)					
SOLE CREASES (bottom of foot)					
TRANSVERSE PALMAR CREASE					
IMPETIGO OR OTHER SKIN INFECTIONS					
OTHER					
HAIR					
SPARSE HAIR					
CURLY SCALP HAIR					
BRITTLE HAIR					
LOW POSTERIOR HAIRLINE					
ABSENCE OF EYEBROWS					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
NORMAL EYEBROWS					
SPARSE EYEBROWS					
BODY HAIR					
OTHER					
LYMPHATIC SYSTEM					
POSTERIOR CERVICAL HYGROMA (nuchal skin)					
LYMPHEDEMA IN LIMBS (build up of lymph fluid)					
CHYLOTHORAX (lymph fluid in lungs)					
OTHER					
MUSCULAR/SKELETAL					
SHORTNESS OF STATURE					
SHORT NECK					
BROAD NECK					
WEBBED NECK					
DEPRESSION OF BREAST BONE (pectus excavatum)					
PIGEON CHEST (pectus carinatum)					
CERVICAL SPINE FUSION					
KYPHOSIS					
SCOLIOSIS					
OSTEOPOROSIS					
JOINT HYPERFLEXIBILITY Small joints- hands and feet Large joints- knees and elbows					
JOINT CONTRACTURES (includes fingers)					
BLUNTLY ENDED FINGERS					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
EXTRA PADDING ON FINGERS AND TOES					
HYPOTONIA (low muscle tone)					
LEG PAIN					
AFO's (ankle foot orthotics)					
OTHER					
NEUROLOGY					
SEIZURES (type)					
AGE OF ONSET					
BRAIN MRI (results)					
EEG (electroencephalogram)					
NCV (nerve conduction studies)					
VENTRICULAR DILATION					
HYDROCEPHALUS					
CORTICAL ATROPHY					
ABNORMAL MYELINATION					
ABSENT CORPUS CALLOSUM					
HEADACHES					
DIZZINESS					
OTHER					
GENITO-URINARY SYSTEMS					
ABNORMALITIES OF PRIMARY SEX ORGANS (ovary/testes)					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
ABNORMALITIES OF EXTERNAL GENITALIA					
CRYPTORCHIDISM (undescended testes)					
ABDOMINAL ULTRASOUND PERFORMED?					
KIDNEY / BLADDER ANOMALIES					
HYPOSPADIAS (misplaced urethra opening)					
CORRECTIVE SURGERY (please specify details in comments column)					
OTHER					
ENDOCRINOLOGY					
DELAYED PUBERTY					
ADVANCED PUBERTY					
LACK OF SEXUAL DEVELOPMENT					
DELAYED BONE AGE					
GROWTH HORMONE TESTING (normal, abnormalities)					
TAKING GROWTH HORMONE					
HYPOGLYCEMIA (low glucose)					
DIABETES (age at onset)					
OTHER					
VISION / EYES					
HYPERTELORISM (wide set eyes)					
PTOSIS (droopy eyelids)					
REFRACTIVE ERRORS (SPECIFY: eg astigmatism; hyperopia; myopia)					
ESOTROPIA / EXOTROPIA (inward and outward turning of the eyes)					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
STRABISMUS (cross-eyed)					
NYSTAGMUS (jerking movement of the eyes)					
CORTICAL BLINDNESS					
OPTIC NERVE HYPOPLASIA					
RETINA TESTING					
CATARACTS					
GLAUCOMA					
OTHER EYE ABNORMALITIES OR ISSUES					
CORRECTIVE SURGERY (please specify details in comments column)					
WEARS CORRECTIVE LENSES					
OTHER					
HEARING / EARS					
LOW SET EARS					
POSTERIOR ROTATION (ears slightly rotated backwards)					
LARGE/THICK HELICES (outer ears)					
INCOMPLETE FOLDING OF EARS					
CHRONIC OTITIS MEDIA (ear infections)					
TUBES INSERTED					
EXTRA SENSITIVE HEARING					
SMALL EAR CANALS					
EXCESS EAR WAX					
HEARING LOSS					
HEARING TESTED					
WEARS HEARING AIDS					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st</i> <i>experienced</i> <i>or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
OTHER					
RESPIRATORY					
RESPIRATORY PROBLEMS(specify details in comments column)					
FREQUENT RESPIRATORY INFECTIONS					
ASTHMA					
LUNG ABNORMALITIES					
OXYGEN NEEDED (why?)					
TRACHEAL MALACIA (small, weak, collapsible trachea)					
OTHER					
GI SYSTEM					
FAILURE TO THRIVE					
NG TUBE FEEDINGS					
G-TUBE FEEDINGS					
NISSEN FUNDOPLICATION					
EATING LESS THAN NORMAL					
EATING MORE THAN NORMAL					
FOOD/TEXTURE AVERSION					
DISTENDED ABDOMEN					
DIGESTIVE/INTESTINAL PROBLEMS					
FREQUENT OR FORCEFUL VOMITING					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
SWALLOWING DIFFICULTIES					
MALROTATION					
CONSTIPATION					
ESOPHAGUS ABNORMALITIES OR PROBLEMS					
NARROWING OF ESOPHAGUS					
OTHER					
MISCELLANEOUS					
TELL US ANYTHING ELSE WE SHOULD KNOW					
SLEEP PATTERNS					
POOR SLEEPING PATTERNS					
NIGHT TERRORS					
LEG CRAMPS DURING SLEEP					
SLEEP WALKING					
SLEEP APNEA					
SLEEP STUDIES (results)					
OTHER					

CHARACTERISTIC	N/A	Do not Know	AGE <i>1st experienced or noticed</i>	Details of any Specialist consulted & Prognosis	COMMENTS Details of Current % Degree or Delay
BEHAVIORAL / SOCIAL					
CHRONIC CRYING					
STUBBORNNESS					
IRRITABILITY					
OBSESSIVE BEHAVIOR					
OVERACTIVITY					
UNDERACTIVITY					
WITHDRAWN OR DEPRESSED					
AGGRESSIVE BEHAVIOR					
SHORT ATTENTION SPAN					
AUTISM					
ENGAGING PERSONALITY					
ATTENTION DEFICIT HYPERACTIVITY DISORDER					
ATTENTION DEFICIT DISORDER					
OTHER					

Medication Information

CONDITION	MEDICATION NAME	AGE STARTED	AGE STOPPED	LENGTH OF TIME USED <i>Months / years</i>	DOSAGE <i>mg / ml</i>	FREQUENCY <i>(Circle/check)</i>
HEART						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
STOMACH						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
GROWTH HORMONE						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
SEIZURES						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
OTHER (Specify condition)						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
OTHER (Specify condition)						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day
OTHER (Specify condition)						<input type="checkbox"/> 1 x day <input type="checkbox"/> 2 x day <input type="checkbox"/> 3 x day

Details of Medical Equipment & Augmentative Devices

MEDICAL EQUIPMENT & DEVICES	NEVER USED	AGE FIRST USED	LENGTH OF TIME USED	COMMENTS & DETAILS
STANDER				
WALKER				
WHEELCHAIR				
HEARING AIDS				
MEDICAL / ADAPTED BED				
AUGMENTATIVE COMMUNICATION DEVICES (SPECIFY DETAILS)				
OTHER				

Developmental Milestone Details

GROSS MOTOR SKILLS MILESTONE	AGE ACHIEVED (Months / Years)	COMMENTS
ROLLED		
CRAWLED		
SAT UP UNASSISTED		
STOOD		
WALKED		
RAN		
JUMPING / HOPPING / BALANCING		
RODE A SCOOTER / BIKE		
OTHER		

Speech Development Information

SPEECH DEVELOPMENT MILESTONE	AGE ACHIEVED (Months / Years)	COMMENTS
VOCALIZATIONS		
BABBLING		
FIRST WORD		
VOCABULARY MORE THAN 10 WORDS		
PUTS 2 OR MORE WORDS TOGETHER		
SPOKEN VOCABULARY (Number of words & current level of vocabulary development)		
SIGN LANGUAGE (Specify type of sign program)		

Intervention Programs Information

TYPE OF PROGRAM	AGE STARTED	COMMENTS Time per session/hours & number of times per week If no longer in program, number of years participating Comment on home activities if relevant
EARLY INTERVENTION		
OCCUPATIONAL THERAPY		
PHYSICAL THERAPY		
SPEECH THERAPY		
SIGN LANGUAGE		



CFC #

Patient Consent for Medical Photography

Patient name: _____ Date: _____

Check here if minor or if patient is unable to provide consent

I consent for medical photographs to be made of me or my child (or person for whom I am legal guardian). I understand that the information may be used in my medical record, for purposes of medical teaching, or for publication in medical textbooks or journals as I have designated below. By consenting to these medical photographs I understand that I will not receive payment from any party. Refusal to consent to photographs will in no way affect the medical care I will receive. If I have any questions or wish to withdraw my consent in the future I may contact:

Brenda Conger, President of CFC International 607-772-9666

By signing **one** of the lines below I confirm that this consent form has been explained to me in terms in which I understand.

- 1) I consent for these photographs to be used in medical publications, including medical journals, textbooks, and electronic publications. I understand that the image may be seen by members of the general public, in addition to scientists and medical researchers that regularly use these publications in their professional education. Although these photographs will be used without identifying information such as my name, I understand that it is possible that someone may recognize me. I also agree for my image to be shown for teaching purposes and to be used for my medical record:

_____ (signature)

- 2) I agree for my image to be shown for teaching purposes **AND** to be used for my medical record, but **NOT FOR** medical publication:

_____ (signature)

- 3) I agree to use my image for medical records **ONLY**:

_____ (signature)

For patients between ages 7 and 18 years, a signature below indicates that the information in this consent form has been explained to me, and I assent to use my images as outlined above:

_____ (signature of patient)

_____ (witness)

CHECKLIST

- Completed registry**
- Photos of individual at different ages**
- Copies of specialist reports**
- Copy of clinical test results**
- Two completed photo consents (one for CFC International on page 3 and one for research on page 22)**