CF screening in adults and transition to adult care

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Since 2003

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I do not have any conflict of interest

I turn the topics around

Transition, definition

The process or a period of changing from one state or condition to another.

dictionary Oxford Languages

Transition in CF care

Changing from paediatric to adult care.

Transition, what are the problems / issues?

- Getting older, end of puberty, insecure, mentally, physically
- Changes in school/profession, friends
- Relationships, choices for the future, family-planning
- Getting independent from parents
- Parents have to let go
- And at the same time, a new medical team
- Maybe some lost of information

Transition, what are the problems / issues?

- Anxiety leaving the pediatric health care which have been the primary care providers for their entire lives. (parents more then child.)

Daniel Office / I. Heeres 2022: Transition from paediatric to adult care in cystic fibrosis

- Transition of CF care from pediatric to an adult based multidisciplinary team is a complex process and careful coordination with a transition key worker is necessary for successful transition without adverse outcome.

Dhochak N, Kabra SK 2023: Transition Care in Cystic Fibrosis

Transition, what are the problems / issues?

- There is evidence that transfer of care for older adolescent patients to adult care is associated with a deterioration in health, especially in those with chronic conditions.

Osborne C, Mannerfeldt J, Brain P, McQuillan SK. 2020: Difficulties in Transition of Care from Pediatric to Adult Gynecology Providers: Should We Maintain Care into Adulthood?

- Good communication and collaboration between pediatric and adult care teams is crucial to ensure a smooth transfer of care.

Willis LD 2020: Transition From Pediatric to Adult Care for Young Adults With Chronic Respiratory Disease

The role of the CF nurse in the CF team

- Some descriptions:
- At a glance, your CF nurse: Helps coordinate and carry out medical care plans. Walks you through your daily treatment plan and any changes. Helps facilitate communication between you and the other members on your CF care team.

CFF.org.uk

- A five-fold responsibility: advocacy, clinical management, advice and support, education, research and management

2001, National Consensus Standards for the Nursing Management of cystic fibrosis CFF.org.uk

The role of the CF nurse in the CF team

The role of the CF Clinical Nurse Specialist should include education, advocacy and psychosocial support, particularly at important times such as:

- notification of a screening result and diagnosis
- first admission to hospital
- first course of intravenous antibiotics
- a second diagnosis (e.g. CF-related diabetes)
- **transition from pediatric to adult care**
- reproductive issues, pre- and postnatal care
- transplant and end-of-life issues

2014 ECFS Standards of Care: Framework for the Cystic Fibrosis Centre

Transfer

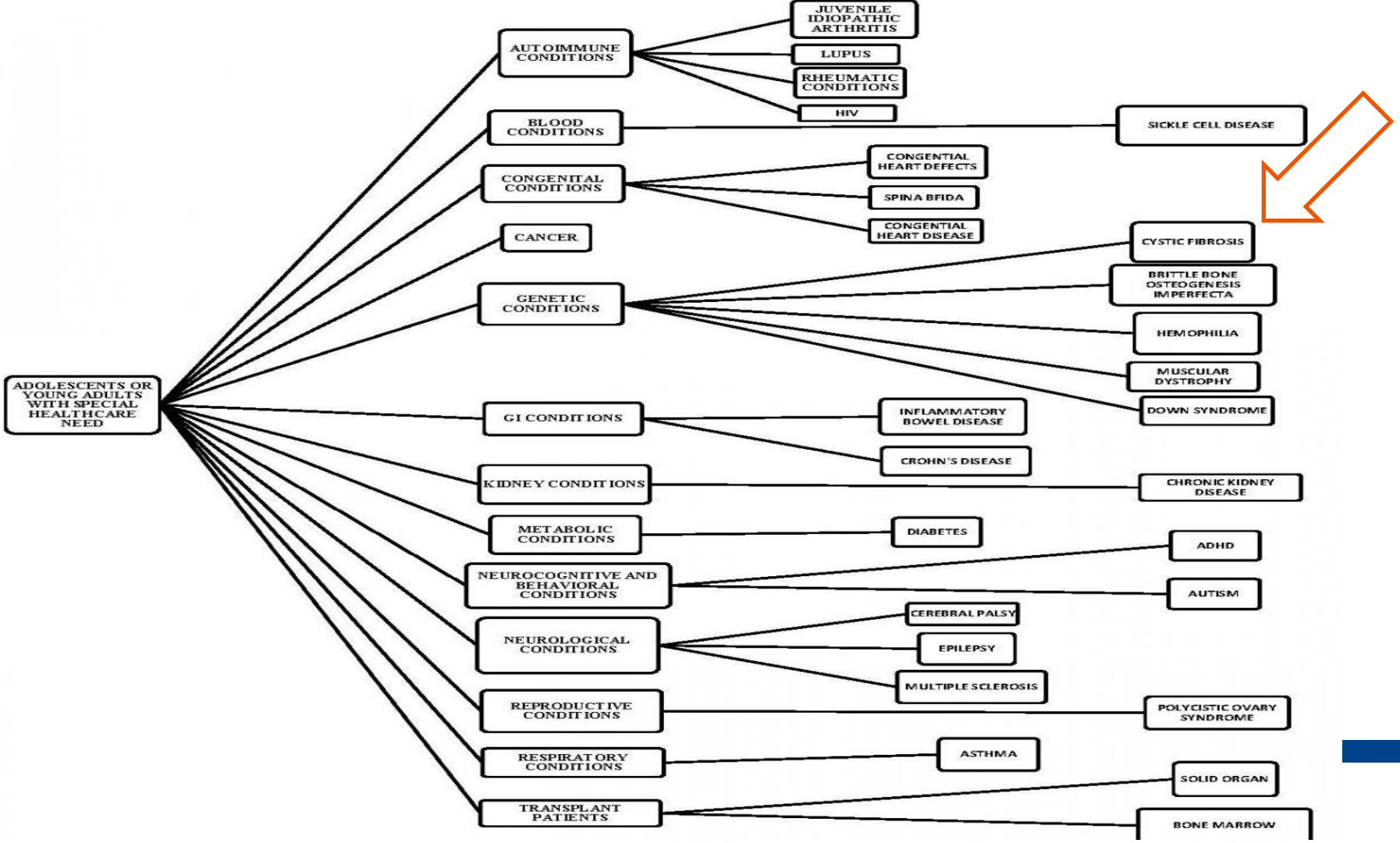
The young person with CF and his/her family must be involved in planning transfer at an early stage. The topic should be introduced when the diagnosis of CF is made and reinforced at appropriate intervals thereafter. Practical discussions should start at around 11 years of age in the context of educational, social and sexual conversations about growing up with a long-term condition.

2014 European Cystic Fibrosis Society Standards of Care: Framework for the Cystic Fibrosis Centre

Transitions of Care From Pediatric to Adult Services for Children With Special Healthcare Needs

Research Protocol Nov 19, 2020

Agency for Healthcare Research and Quality


ADOLESCENTS OR YOUNG ADULTS WITH SPECIAL HEALTHCARE NEED
AUTOIMMUNE CONDITIONS
JUVENILE IDIOPATHIC ARTHRITIS
LUPUS
RHEUMATIC CONDITIONS
HIV
BLOOD CONDITIONS
SICKLE CELL DISEASE
CONGENITAL CONDITIONS
CONGENTIAL HEART DEFECTS
SPINA BFIDA
CONGENTIAL HEART DISEASE
CANCER
GENETIC CONDITIONS
CYSTIC FIBROSIS
BRITTLE BONE OSTEOGENESIS IMPERFECTA
HEMOPHILIA
MUSCULAR DYSTROPHY
DOWN SYNDROME
GI CONDITIONS
INFLAMMATORY BOWEL DISEASE
CROHN'S DISEASE
KIDNEY CONDITIONS
CHRONIC KIDNEY DISEASE
METABOLIC CONDITIONS
DIABETES
NEUROCOGNITIVE AND BEHAVIORAL CONDITIONS
ADHD
AUTISM
NEUROLOGICAL CONDITIONS
CEREBRAL PALSY
EPILEPSY
MULTIPLE SCLEROSIS
REPRODUCTIVE CONDITIONS
POLYCISTIC OVARY SYNDROME
RESPIRATORY CONDITIONS
ASTHMA
TRANSPLANT PATIENTS
SOLID ORGAN
BONE MARROW

Main points: Executive Summary

- With only a single exception that showed no benefit, we found that for all outcomes and interventions the evidence was insufficient to draw conclusions because the uncertainty of evidence was too high. Insufficient evidence does not mean that the intervention is of no value to children with special healthcare needs (CSHCN).
- Transition clinics may not improve hemoglobin A1C levels at 12 or 24 months in youth with type 1 diabetes mellitus compared with youth who received usual care (low-strength evidence).
- While significant barriers impede implementation of interventions, some approaches to reduce these barriers in future interventions include dedicating time and resources to support transition planning, developing a workforce trained to care for the needs of this population, and creating structured processes and tools to facilitate the transition process.
- No globally accepted definition exists for effective transition of care from pediatric to adult services for CSHCN, nor is a single measure or set of measures consistently used to evaluate effectiveness of transitions of care.
- The literature identifies only a limited number of available trainings or other implementation strategies, generally focused on specific clinical specialties in targeted settings.
- No included studies measured the effectiveness of providing linguistically and culturally competent healthcare for CSHCN.
- Trainings and interventions to prepare pediatric patients and their families for transitioning CSHCN to adult care vary considerably in their components, structures, and processes.

Main points

- No globally accepted definition exists for effective transition of care from pediatric to adult services for CSHCN, nor is a single measure or set of measures consistently used to evaluate effectiveness of transitions of care.

After all these years, why??

Main points

- While significant barriers impede implementation of interventions, some approaches to reduce these barriers in future interventions include dedicating time and resources to support transition planning, developing a workforce trained to care for the needs of this population, and creating structured processes and tools to facilitate the transition process.

So, it needs time, money and effort.

Main points

- The literature identifies only a limited number of available trainings or other implementation strategies, generally focused on specific clinical specialties in targeted settings.

- Trainings and interventions to prepare pediatric patients and their families for transitioning CSHCN to adult care vary considerably in their components, structures, and processes.

So, where do we find good training / tools ?

The role of the CF nurse in transition

Both the pediatric and adult CF Clinical Nurse Specialists play an important role in ensuring a successful transition and will manage details such as: patient and parent involvement in decision making; clear communication between pediatric and adult CF MDTs; appropriate transition clinics involving the MDT; ensuring attendance at the adult clinic with appropriate follow-up.

2014 ECFS Standards of Care: Framework for the Cystic Fibrosis Centre

Transition, what do I need as patient / parent?

- A good preparation,
- Explanation about the new team,
- See the new faces, places,
- Explanation about changes in communication,
- Gain confidence in the new team
- Knowing all I have to know about my illness.

Transition, what do I need as CF-nurse?

- Good planning
- Time
- Patient confidence
- Parents confidence
- Protocols / guidelines

Transition, protocols tools

An example:

Pediatric to Adult Care Transitions Tools ` Transition Readiness Assessment for Parents/Caregivers of Youth with Physical Disabilities

Pediatric to Adult Care Transitions Initiative

ACP, American College of Physisians

Please fill out this form to help us see what you already know about your health and how to use health care and the areas that you need to learn more about. If you need help completing this form, please ask your parent/caregiver.

Date:

Name: Date of Birth:

Transition Importance and Confidence *On a scale of 0 to 10, please circle the number that best describes how you feel right now.*

How important is it to you to prepare for/change to an adult doctor before age 22?

0 (not)	1	2	3	4	5	6	7	8	9	10 (very)

How confident do you feel about your ability to prepare for/change to an adult doctor?

0 (not)	1	2	3	4	5	6	7	8	9	10 (very)

My Health *Please check the box that applies to you right now.*	*Yes, I know this*	*I need to learn*	*Someone needs to do this... Who?*
I know my medical needs.	☐	☐	☐
I can tell others what my medical needs are.	☐	☐	☐
I know my symptoms including ones that I quickly need to see a doctor for.	☐	☐	☐
I know what to do if I have a medical emergency.	☐	☐	☐
I know who to contact in my family or among friends if I have a medical emergency.	☐	☐	☐
I know my medications and what they are for.	☐	☐	☐
I know when to take my medications without a reminder.	☐	☐	☐
I know my allergies.	☐	☐	☐
I know what do in case of an allergic reaction.	☐	☐	☐
I know my non medication treatments (for example occupational and physical therapy) and when I need to do them.	☐	☐	☐
I know my assistive devices.	☐	☐	☐
I know how to maintain my assistive devices.	☐	☐	☐

Using Health Care	*Please check the box that applies to you right now.*	*Yes, I know this*	*I need to learn*	*Someone needs to do this… Who?*
I know who my doctors are.		☐	☐	☐
I know how to tell my doctor's office about the special assistance I need prior to my first visit (e.g., help with transferring)		☐	☐	☐
I know how I can reach my doctor (phone, email, text, etc.)		☐	☐	☐
I make and keep track of my own doctor appointments.		☐	☐	☐
Before a visit, I think about questions to ask about my health.		☐	☐	☐
I have a way to get to my doctor's office.		☐	☐	☐
I know to show up 15 minutes before the visit to check in.		☐	☐	☐
I know where to go to get medical care when the doctor's office is closed.		☐	☐	☐
I have a file/folder at home for my medical information.		☐	☐	☐
I have a copy of my medical summary and emergency care plan.		☐	☐	☐
I have a copy of my plan of care.		☐	☐	☐
I carry important health information with me every day (e.g. insurance card, allergies, medications, emergency contact information, and medical summary).		☐	☐	☐
I know how to fill out medical forms.		☐	☐	☐

Pediatric to Adult Care Transitions Tools

Transition Readiness Assessment for *Youth with Physical Disabilities*

I know how to ask for a form to be seen by other doctors/therapists.	☐	☐	☐
I know where my pharmacy is and what to do when I run out of my medication.	☐	☐	☐
I know where to get blood tests or x-rays if my doctor orders them.	☐	☐	☐
I understand how health care privacy changes once I turn 18.	☐	☐	☐
I have a plan so I can keep my health insurance after age 18	☐	☐	☐
My family and I have discussed my ability to make my own health care decisions at age 18.	☐	☐	☐

Tools

- Ready, steady, go,
- CF Myway,
- “op eigen benen”: Dutch platform

When should transition begin and end?

- Start around 10-11 years: preparation
- Yearly questionaries
- Actual transition 18 (maybe question of insurance)
- End transition around 24: evaluation.

One CF nurse for paediatric and adult

Situation in my CF centre (CF centre Maastricht):

- 2 CF nurses, both responsible for pediatric **and** adult patient.
- Nurses are present at all medical visits of the patients.
- Last pediatric visit the pulmonologist gets acquainted (CF nurse organizes this)
- Transition after the yearly check-up, so all the latest results are known.

The benefit of one nurse

- The way to contact the CF team is the same (we are the first point of contact for the patients)
- No information is lost
- Good connection between both team:
    - More of less the same working method
- Patients do not have to let go of a long-established relationship
- CF nurse can inform the adult doctor about a personal approach of the patient

Transition, patient's experiences

- I have experienced little difference from the transition, which I experience as positive.
- I was a little stressed about it, but discovered that a number of things remained the same.
- I was afraid that I could not sufficiently explain my situation, but my CF nurse helped me with that.

Transition parent's experiences

- I thought the transition was well organized and we received concrete explanations about it at an early stage.
- It felt good as a mother to leave some of the control to my adult sons and slowly let them take on their own responsibility (with the adult CF team as backup).
- I must say that the trusted point of contact (CF nurse) made a big positive difference for them.

Questions?

- Do you work with specific tools?
- Who works also with the same CF nurse for pediatric and adult team?

Late diagnosis, definition

A late diagnosis is defined as an individual above the age of 18 years who is found to have cystic fibrosis

www.cysticfibrosis.ca

Late diagnosis adults

Research has shown that people with late diagnosis demonstrate fewer complications, less serious lung disease, and a lower incidence of pancreatic problems.

www.cysticfibrosis.ca

Most common rare mutation:

- R177H
- Male with infertility
- Generally mild CF.
- Very individual approach.

Late diagnosis, children

- Unexpected because of neonatal screening.
- NLD: since 2011 screening
- Negative screening possible with severe meconium ileus.
- Other cases very rare

Case, girl 7 year

- Normal birth,
- No meconium ileus
- Neonatal screening neg
- Since 2nd year of life pulmonary problems

Symptoms since 2nd year of life

- Chronic coughing
- Mucus
- Slightly increased infection parameters
- Normal growth

Differential Diagnosis?

- Adenoid-hypertrophy
- Hyperreactivity
- Dysfunctional breathing
- Reflux (GERD)

Diagnostics in own hospital

- Blood test: just increased infection
- X-ray of Oesophagus (swallow Barium): negative
- Pulmonary CT scan: extensive bronchiectasis
- No sweat test, because of negative neonatal screening

Treatment

- Flixotide
- Mometason nose-spray
- Nexium
- In case of extreme effort: prednisone (swimming exam)

Second opinion MUMC (paediatric pulmonologist)

- Lung function test: 106% FEV1
- Sputum culture: Hemophilus influenzae
- Sweat test: 72 Mmol/L
- Elastase: >500 ug/g
- DNA: delta F508 / A544E

Treatment CF center

- Kaftrio / Kalydeco
- Hypertonic saline nebulization
- Azithromycin
- Physiotherapy

Physical effect

- Less sputum
- Less coughing (only with nebulization)
- Less stomach pain
- With exacerbation direct antibiotics with good effect

Mental effect

- Relieve for child and parents
- Told directly at school about her CF
- But also feelings of guild for parents (father = general doctor)
- After 5 months some stress returned
- School results now lower
- Difficulty falling asleep
- Nebulization gives stress

Questions?

A very special case: adult

- Female, 42 years. No pulmonary problems, just some asthma.
- 2014 daughter, neonatal screening CF positive
- Mutations daughter: Delta F508 / G628R (UV)
- CFTR2 database:
    - **This variant combination causes CF.**
    - **Patients with CF who have this variant are likely to be pancreatic sufficient.**
    - There are 9 patients with this variant combination in the CFTR2 database.

Daughter (2014)

- Sweat test: 63 mmol/L chloride
- Elastase: >500 ug/g
- Lung function tests 2019 until now: FEV1 >100%, except 1x 68%: exacerbation – Augmentin.
- Pulmonary CT-scan: some peri-bronchial thickening. No mucus plugging
- No medication, No Kaftrio (parents choice)

Genetic counseling

Father: delta F508

Mother: G628R (UV)

Feelings of guilt and worries about the future ⟶ psychological counseling.

Possibilities for next pregnancy: PNS or PGD (permission has to be asked)

2nd pregnancy 2016

10 weeks PNS: Chorionic Villus Sampling (CVS)

12 weeks result: Fetus is carrier of delta F508, no G628R

21 weeks ultrasound: intestinal loop dilatation, suspicious for CF

2nd pregnancy 2016

Amniocentesis with full CF-sequencing

Also full CF-sequencing in blood both parents

22+5 weeks: mother has also G542X mutation. At 2 allels.
Also G542x found in amnion.

2 nd Pregnancy 2016

Mutation delta F508 / G542x in CFTR2 database:

- **This variant combination causes CF.**
- **This variant combination is expected to cause pancreatic insufficiency.**
- There are 2,112 patients with this variant combination in the CFTR2 database.

Combination of these 2 mutations causes severe CF.

23+2 weeks: termination of pregnancy

Follow-up

- Psychological support for both parents:
  - Loss of a baby in an advanced pregnancy
  - Realization mother of having also CF
  - Feelings of guilt for giving her children CF
  - How about a second child?

How about genetic testing?

- Parents of CF child: only searching for mutation known at child.
- In this case the genetic test was just looking for the delta F508 and the G628R
- Reason why other mutation in mother and fetus was not found.
- (Dutch policy?)

CF counseling mother

By CF nurse shortcut to pulmonologist (because same nurse for pediatric and adults).

No need to consult another person /explain her situation.

Advice pulmonologist: a total check-up

Follow-up

In 2020 little brother, through PGD, third attempt.

2024 mother was ready to make an appointment pulmonologist

Partiel Check-up

- Sweat test: 99 mmol/L chloride
- Elastase: >500 ug/g
- Lung function test: FEV1: 91%
- CT scan: Some bronchiectasis, peribronchial thickening, mucusplugging
- Medication: start nebulization with hypertonic salin.
- Physiotherapy: mucus evacuation / exercises

Follow-up

Questions from daughter (now 9 years old):

- Do we have the same disease?
- Do I have to nebulize too?
- Do I have to do also physiotherapy?

Acceptance for mother and daughter seems getting easier, because they can do it together / they share the same situation.

Questions?

- What went wrong? Could this have been prevented?
- What could we do as a CF nurse?
- What counseling could be offered at this family?
- What about the late diagnosis for mother?