

CCHS Survival Guide: Transitioning to Adult Care The Fun Facts You Need

**CCHS Network Family Conference
June 28, 2024**

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Meet the Campbell Family!

- Riley is 22 year old
- Mask/vent dependent during sleep; on a Trilogy
- Trying to establish care in the adult world



Objectives

1. General transition concepts
2. CCHS specific transitional care
3. Medical issues in the CCHS adolescent/young adult



Context

All youth, regardless of their individual challenges, are provided supportive relationships across all transition domains.

Healthcare Transition

“The purposeful, planned movement of adolescents and young adults with chronic physical and medical conditions or intellectual or developmental disabilities from child-centered to adult-oriented healthcare in a way that considers typical developmental processes at this stage of life as well as access, appropriateness and continuity of health care services.”

Connection to cross domain services, information, and supports that prepare them to:

- *Make informed choices about their lives*
- *Foster self-determination and planning*
- *Prepare for self-healthcare management*
- *Pursue education, training, or employment*
- *Be financially literate and independent*
- *Develop leadership and advocacy skills*

Figure 1. N-PeRC healthcare transition definition.

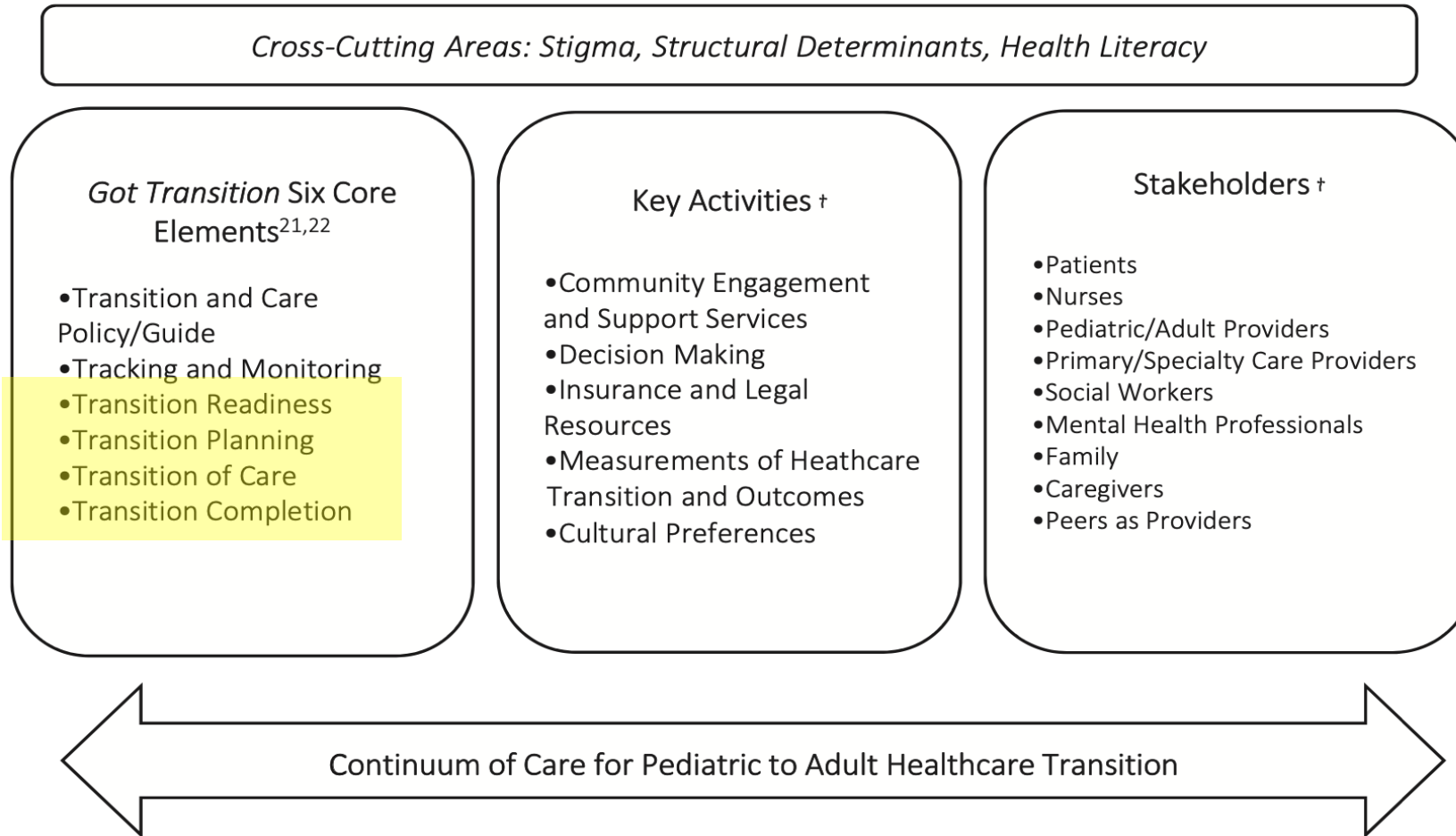


Figure 3. Disease inclusive cross-cutting framework for healthcare transition. †Order of key activities and stakeholders is not representative of importance.

Side-by-Side Comparison

The Six Core Elements of Health Care Transition™ 3.0 are intended for use by pediatric, family medicine, med-peds, and internal medicine practices to assist youth and young adults as they transition to adult-centered care. They are aligned with the AAP/AAFP/ACP Clinical Report on Health Care Transition.¹ Sample tools, implementation guidance, measurement, and payment resources are available at www.GotTransition.org.

**TRANSITIONING YOUTH TO AN ADULT
HEALTH CARE CLINICIAN**
*(For use by Pediatric, Family Medicine, and Med-Peds
Clinicians)*

1. Transition and Care Policy/Guide

- Develop a transition and care policy/guide with input from youth and parents/caregivers that describes the practice's approach to transition, an adult approach to care in terms of privacy and consent, and age of transfer to an adult clinician.
- Educate all staff about the practice's approach to transition and distinct roles of the youth, parent/caregiver, and pediatric and adult health care team in the transition process, taking into account cultural preferences.
- Display transition and care policy/guide somewhere accessible in practice space, discuss and share with youth and parent/caregiver, beginning at age 12 to 14, and regularly review as part of ongoing care.

2. Tracking and Monitoring

- Establish criteria and process for identifying transition-aged youth.
- Develop process to track receipt of the Six Core Elements, integrating with electronic medical records (EMR) when possible.



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3. Transition Readiness

- Conduct regular transition readiness assessments, beginning at age 14 to 16, to identify and discuss with youth and parent/caregiver their needs for self-care and how to use health care services.
- Offer education and resources on needed skills identified through the transition readiness assessment.



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.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can tell others what my medical needs are.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know my symptoms including ones that I quickly need to see a doctor for.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know what to do if I have a medical emergency.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know who to contact in my family or among friends if I have a medical emergency.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know my medications and what they are for.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know when to take my medications without a reminder.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know my allergies.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know what do in case of an allergic reaction.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know my non medication treatments (for example occupational and physical therapy) and when I need to do them.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know my assistive devices.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know how to maintain my assistive devices.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
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.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
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)		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know how I can reach my doctor (phone, email, text, etc.)		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I make and keep track of my own doctor appointments.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Before a visit, I think about questions to ask about my health.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have a way to get to my doctor's office.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know to show up 15 minutes before the visit to check in.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know where to go to get medical care when the doctor's office is closed.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have a file/folder at home for my medical information.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have a copy of my medical summary and emergency care plan.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have a copy of my plan of care.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I carry important health information with me every day (e.g. insurance card, allergies, medications, emergency contact information, and medical summary).		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know how to fill out medical forms.		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

From Boston Children's



● ARE YOU READY TO TRANSITION?

1 CAN YOU DESCRIBE YOUR **MEDICAL CONDITION** TO SOMEONE ELSE?

2 DO YOU KNOW THE MEDICATION AND/OR EQUIPMENT YOU USE TO **MANAGE**

● **YOUR CONDITION?**

3 DO YOU **RELY ON YOUR PARENTS** TO SPEAK FOR YOU WHILE MEETING WITH DOCTORS?

4 CAN YOU **SET UP AN APPOINTMENT** BY YOURSELF?

5 DO YOU UNDERSTAND **HOW INSURANCE**

● **WORKS** AND HOW IT AFFECTS YOU?



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Side-by-Side Comparison

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4. Transition Planning

- Develop and regularly update the plan of care, including readiness assessment findings, youth's goals and prioritized actions, medical summary and emergency care plan, and, if needed, a condition fact sheet and legal documents.
- Prepare youth and parent/caregiver for an adult approach to care, including legal changes in decision-making and privacy and consent, self-advocacy, and access to information.
- Determine need for decision-making supports for youth and make referrals to legal resources.
- Plan with youth and parent/caregiver for optimal timing of transfer from pediatric to adult care. If both primary and subspecialty care are involved, discuss optimal timing for each.
- Assist youth in identifying an adult clinician(s) and provide linkages to insurance resources, self-care management information, and community support services.
- Obtain consent from youth/parent/caregiver for release of medical information.
- Take cultural preferences into account throughout transition planning.

TRANSITIONING YOUTH TO AN ADULT HEALTH CARE CLINICIAN

(For use by Pediatric, Family Medicine, and Med-Peds Clinicians)

Assemble your adult-care team:

- primary care physician
- pulmonologist specializing in ventilators, cardiologist,
- gastroenterologist,
- and sleep medicine physician

Other adult skills to gain

- Understand health insurance coverage, knowledge of accepted providers and hospitals and programs
- Identify and advocate for accommodations needed for success at school/work



State-by-State Variability in Adolescent Privacy Laws

Marianne Sharko, MD, MS,^a Rachael Jameson,^b Jessica S. Ancker, PhD, MPH,^c Lisa Krams, MS,^d Emily C. Webber, MD,^{e,f}
S. Trent Rosenbloom, MD, MPH^c

RESULTS: We observed notable state-by-state variability in laws governing consent for adolescent patients. No states had identical policies for all services studied. For example, although all states had provisions for consent to management of sexually transmitted infections, there were variable specifications in the age and type of minor, whether this includes human immunodeficiency viruses, and whether confidentiality is protected. Providing confidential care to the adolescent patient has been set as a priority by medical societies; however, guidelines are limited by the need to comply with state laws and regulations.

CONCLUSIONS: State laws on consent and privacy for adolescents are highly variable, and many do not reflect pediatric professional standards of care. This inconsistency is a barrier to operationalizing a consistent and equitable experience providing evidence-based medical care and ensuring adolescent privacy protection.

To cite: Sharko M, Jameson R, Ancker JS, et al. State-by-State Variability in Adolescent Privacy Laws. *Pediatrics*. 2022;149(6):e2021053458



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5. Transfer of Care

- Complete transfer package, including final transition readiness assessment, plan of care with transition goals and prioritized actions, medical summary and emergency care plan, and, if needed, legal documents, condition fact sheet, and additional clinical records.
- Confirm date of first adult clinician appointment.
- Prepare letter with transfer package, send to adult clinician, and confirm adult clinician's receipt of transfer package.
- Communicate with selected adult clinician about pending transfer of care.
- Confirm the pediatric clinician's responsibility for care until youth/young adult is seen by an adult clinician.
- Transfer youth/young adult when their condition is as stable as possible.

TRANSITIONING YOUTH TO AN ADULT HEALTH CARE CLINICIAN
(For use by Pediatric, Family Medicine, and Med-Peds Clinicians)

GETTING ORGANIZED BEFORE YOUR FIRST APPOINTMENT

IT MAY BE HELPFUL TO HAVE A WRITTEN LIST OF THINGS YOU WANT TO COVER DURING YOUR APPOINTMENT, BUT DON'T FEEL LIKE YOU NEED TO STICK TO A SCHEDULE.

HELLO

my name is

- LIST OF MEDICATIONS YOU ARE TAKING
- ANY SUPPLIES/EQUIPMENT YOU USE
- ANY URGENT OR MAJOR CONCERNS YOU MAY HAVE GOING ON (FOR EXAMPLE: HOW TO GET REFILLS OF MEDICINE, SUPPLIES SENT TO YOU, ETC.)
- BASIC OUTLINE OF YOUR MEDICAL HISTORY
- HOW YOU BEST LEARN INFORMATION



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6. Transfer Completion

- Contact youth/young adult and parent/caregiver 3 to 6 months after last pediatric visit to confirm attendance at first adult appointment.
- Elicit anonymous feedback from youth/young adult and their parent/caregiver on their experience with the transition process.
- Communicate with adult practice confirming completion of transfer and offer consultation assistance, as needed.
- Build ongoing and collaborative partnerships with adult primary and specialty care clinicians.



Objectives

1. General transition concepts
2. CCHS specific transitional care
3. Medical issues in the CCHS adolescent/young adult



CCHS-specific transitional care

Clinical Autonomic Research

<https://doi.org/10.1007/s10286-022-00908-8>

REVIEW ARTICLE

Transitional care and clinical management of adolescents, young adults, and suspected new adult patients with congenital central hypoventilation syndrome

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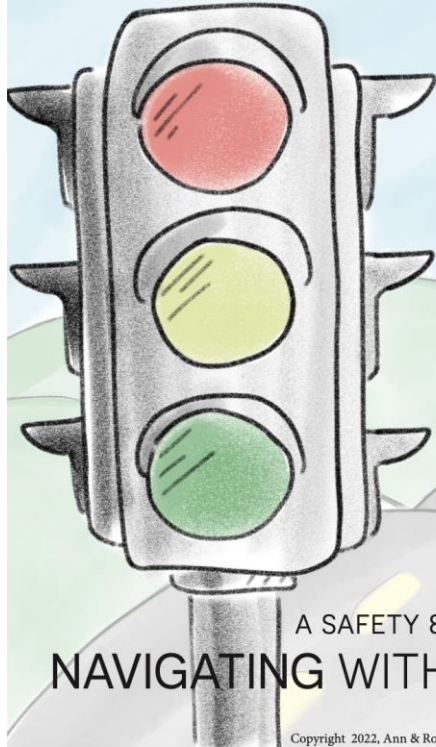


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Self-care and Self-knowledge

ROAD TO INDEPENDENCE



EXTERNAL SUPPORT

INTERNAL SUPPORT

SELF SUPPORT

A SAFETY & WELL-BEING GUIDE FOR NAVIGATING WITH RARE DISEASE

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SELF **SUPPORT**

WHAT'S YOUR BASELINE?

HEART RATE
 AWAKE: _____
 ASLEEP: _____

CO₂ & O₂ LEVELS
 CO₂: _____
 SpO₂: _____

BLOOD PRESSURE
 AWAKE: _____
 ASLEEP: _____

TEMPERATURE

HOURS OF SLEEP _____

HOW CAN YOU MAINTAIN IT?

WHAT SYMPTOMS TELL YOU YOU'RE OFF YOUR BASELINE?

MEDICATIONS & EQUIPMENT TO CARRY AT ALL TIMES?

WHAT ARE YOUR STRESSORS AWAY FROM BASELINE?
HOMEWORK? GRADES? EXTRA-CURRICULAR ACTIVITIES? TIME MANAGEMENT? RESPONSIBILITIES? SELF-CARE? OTHER?

HOW WILL YOU MANAGE THEM?

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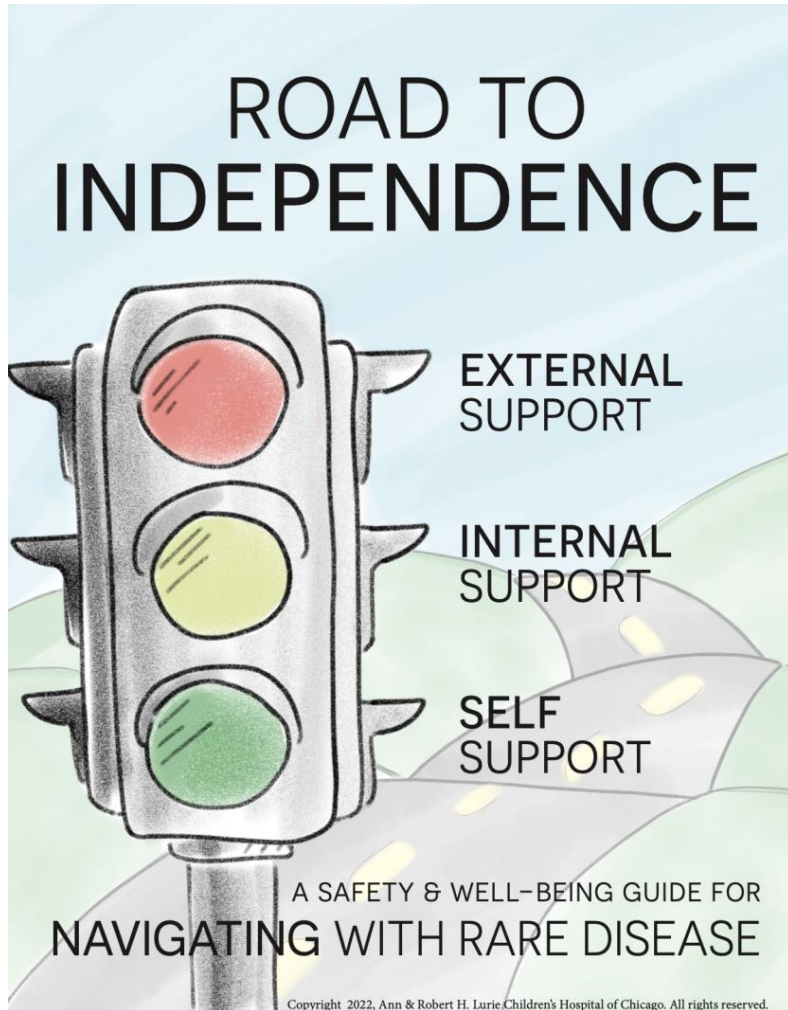
- gradually administer self-care with both medical knowledge and medical skills
- practice conversations in medical planning with family and friends
- to participate in care
- to contribute to emergency plans.
- clear communication of their medical condition and care plans by adults with CCHS to other responsible adults is critical to the maintenance and success of their independence and health



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Internal support



INTERNAL SUPPORT

WHO'S YOUR SUPPORT SYSTEM?

NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____
NAME: _____	PHONE #: _____

PARENTS? SIBLINGS?
COUNSELOR?
TEACHERS? THERAPIST?
FRIENDS? CLASSMATES?
NURSES? RELATIVES?
COUSINS? PETS?

CHECKING IN WITH THEM

WHEN? _____ HOW OFTEN? _____

TALKING POINTS

WHAT SHOULD THEY KNOW?	HOW CAN THEY SUPPORT YOU?
WHAT QUESTIONS MIGHT THEY ASK? HOW CAN YOU RESPOND?	

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The worksheet is titled 'INTERNAL SUPPORT' and features a traffic light illustration at the top. Below the title is a section for identifying support system members, including a table for names and phone numbers. To the right of this table are various categories of support system members with checkboxes: PARENTS?, SIBLINGS?, COUNSELOR?, TEACHERS?, THERAPIST?, FRIENDS?, CLASSMATES?, NURSES?, RELATIVES?, COUSINS?, and PETS?. Below this is a section for 'CHECKING IN WITH THEM' with fields for 'WHEN?' and 'HOW OFTEN?'. The final section is 'TALKING POINTS', which is divided into two columns: 'WHAT SHOULD THEY KNOW?' and 'HOW CAN THEY SUPPORT YOU?', and a row for 'WHAT QUESTIONS MIGHT THEY ASK? HOW CAN YOU RESPOND?'.

- Identify members of the “inner circle”
 - Buddy system
- Create a weekly schedule
- Technology
 - pre-set reminders on a smart phone and watch for sats, CO2, vent initiation, pacers to.
 - wearable options to detect oxygen saturation and heart rate are available.
 - Smarter/smaller ventilators



External Support

EXTERNAL

IND

NAVIGA

CONTACT INFO:

EMERGENCY CONTACT (GENERAL)
 NAME: _____ PF
 ADDRESS: _____

EMERGENCY CONTACT (LOCAL)
 NAME: _____ PF
 ADDRESS: _____

NEAREST HOSPITAL (EMERGENCY ROOM)
 NAME: _____ PF
 ADDRESS: _____

YOUR DOCTOR'S OFFICE
 NAME: _____ PF
 ADDRESS: _____

YOUR DME COMPANY
 NAME: _____ PF
 ADDRESS: _____

FIRE STATION
 NAME: _____ PF
 ADDRESS: _____

EMS SERVICES
 NAME: _____ PF
 ADDRESS: _____

OTHER _____
 NAME: _____ PF
 ADDRESS: _____

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WHAT ARE SOME SIGNS THAT YOU MIGHT NEED TO CONTACT YOUR EXTERNAL SUPPORT SYSTEM?

WHAT DO YOUR CONTACTS NEED TO KNOW TO HELP YOU?

YOUR BASIC EMERGENCY PLAN

- CALL 911
- CALL BOTH OF YOUR EMERGENCY CONTACTS
- GET TO THE NEAREST EMERGENCY ROOM

ANY ADDITIONAL STEPS?

ADDITIONAL HEALTH INFO?

PERSONAL INFO

NAME: _____
 PHONE #: _____
 ADDRESS: _____
 DATE OF BIRTH: _____
 GENDER: _____ RACE: _____

CARDIAC PACEMAKER?	YES	NO
DIAPHRAGM PACEMAKER?	YES	NO
VENTILATOR?	YES	NO
TRACH?	YES	NO

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- Portable Medical Health Summary readily available -- digital
- It is also advisable for each patient with CCHS to carry critical medical information available on a smart watch, medical bracelet or necklace, or card located on a phone cover



Portable Medical Summary

Preferred name: _____
Legal name: _____
Pronouns: _____
Date of birth: _____

Preferred language or communication method:

Main diagnoses

Dates and comments

Key medical history (e.g., treatments, surgeries, procedures that are relevant today)

Dates and comments

Medications

Purpose/reason to take

Dose & frequency

Medications	Purpose/reason to take	Dose & frequency

Medical devices: _____

Allergies/reactions: _____

Medical equipment & supplies

Provider

Contact info

--	--	--

Nutrition supplies

Provider

Contact info

--	--	--

Important related issues and risks: (What to watch for in my healthcare, due to my past conditions and treatment, e.g., long-term effects of chemo, susceptibility to certain infections, etc.)

Portable Medical Summary

Name: _____ Date of birth: _____
Address: _____ Phone: _____
Email: _____

My baseline is: (Behavioral, neurological, communication ability, etc.)

For my healthcare visits & interactions, I need you to know:
Anything else a provider should know to support your care experience

Home situation: (Live with family, college dorm, roommates, group home, etc.)

Emergency/family contacts

Phone

Relationship (parent, guardian etc.)

--	--	--

Healthcare providers

Phone

Specialty/reason seen

Insurance information: Complete this section or attach a copy of the front and back of your insurance card.

Insurance company/plan: _____ ID number: _____

Name of policy holder: _____ Group number: _____

Contact phone for providers: _____ Contact phone for members: _____

Lifestyle information: (School, job, community activities, athletics, hobbies, etc.)

Things I need from my medical team:

Objectives

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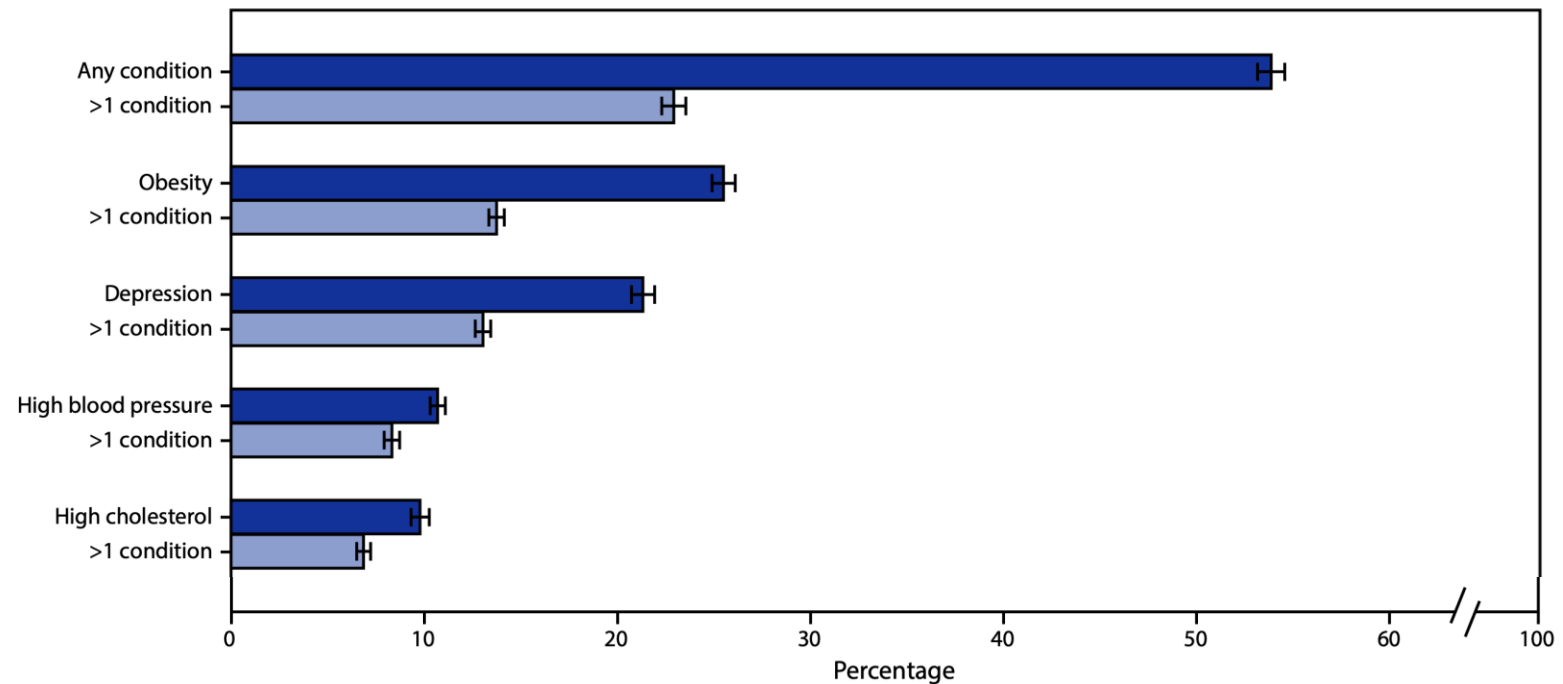
Common conditions are common



Chronic Conditions Among Adults Aged 18–34 Years — United States, 2019

Weekly / July 29, 2022 / 71(30);964–970

FIGURE 1. Percentage* of chronic conditions† among adults aged 18–34 years — Behavioral Risk Factor Surveillance System, United States, 2019



Ventilatory Management

Clinical findings	Management Approaches
<ul style="list-style-type: none"> ▪ Absent ventilatory responses, unreliable arousal to low O₂/high CO₂ <ul style="list-style-type: none"> ▪ Can associate headaches, fatigue with <u>hypovent</u> ▪ Alveolar hypoventilation, worsened during sleep, NREM>REM ▪ Shallow breathing with monotonous respiratory rate, some central apneas ▪ Adequate ventilation while awake in 75% ▪ Can develop OSA ▪ Severe breath-holding spells in infants/toddlers 	<ul style="list-style-type: none"> ▪ Lifelong mechanical assisted ventilation during sleep; variable during wake ▪ Aggressive vent support, especially in early childhood ▪ Titration of support at least annually, more frequently during early childhood, times of rapid growth, or instability <ul style="list-style-type: none"> ▪ Goal SpO₂>95% with FiO₂ <40% ▪ Goal PCO₂ 30-45 mmHg ▪ Increase support with illnesses, sedation ▪ Critical need for objective monitoring <ul style="list-style-type: none"> ▪ Oximetry during sleep ▪ PPV with disconnect alarms ▪ Capnography for spot checks ▪ Supervision/sponsor/support <ul style="list-style-type: none"> ▪ E.g., swimming <p>Childhood</p> <ul style="list-style-type: none"> ▪ Transition from PPV/trach as infant to PPV/mask or DP at school age <ul style="list-style-type: none"> ▪ Monitor mid-face with early PPV/mask ▪ DP does not guarantee decannulation ▪ Nursing support

- “you have one job” = use the ventilator
- Identify a pulmonary or sleep physician who if not familiar with CCHS, willing to listen and learn!
- Sleep lab that incorporates capnography (CO₂ monitoring); annual sleep study
- “Adult” DME if not already transitioned
- Copies of letters, justifications, appeals written for equipment to date
- Determine what is needed for exercise (next slide)
- Ensure equipment and settings are updated before transition (as close to transition as possible)
- Annual labs
- Nightly oximetry; screening oximetry
- Decannulate/place pacers prior to transition
- Development of obesity-related OSA
- Address craniofacial issues from prolonged mask use prior to transition



Exercise Management

TABLE 3 Comparison of nadir oxygen saturation, peak end-tidal carbon dioxide and predicted 6MWD based on *PHOX2B* genotype.

Variable	20/25 and 20/26 PARM, <i>n</i> = 7 ^a	20/27 and 13/27 PARM, <i>n</i> = 6 ^a	NPARM, <i>n</i> = 2 ^a	<i>p</i> Value ^b
Nadir SpO ₂ (%)	92 (91–92.5)	90 (90–90)	93.5 (93.2–93.8)	0.029
Peak ETCO ₂ (mmHg)	32 (30–37)	49 (42–52.2)	32.5 (26.2– 38.8)	0.046
Predicted 6MWD (%)	61.6 (51.2–64.2)	58.3 (51.9–59.9)	59 (49.4–68.6)	0.8

Abbreviations: 6MWD, 6-min walk distance; ETCO₂, end-tidal carbon dioxide; NPARM, nonpolyalanine repeat mutation; PARM, polyalanine repeat mutation; *PHOX2B*, paired-like homeobox; SpO₂, oxygen saturation.

^aMedian (interquartile range).

^b*p* Value based on Kruskal–Wallis rank sum test.

Impaired ventilation during 6-min walk test in congenital central hypoventilation syndrome

Radhika N. Ghosh MD¹ | Lokesh Guglani MD¹ | Adrianna L. Westbrook MPH² |
Chad Y. Mao MD³ | Shasha Bai PhD² | Thomas G. Keens MD⁴ | Ajay S. Kasi MD¹

Pediatric Pulmonology. 2022;1–8.



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Cardiac Management

Clinical findings	Management Approaches
<ul style="list-style-type: none"> Bradyarrhythmia Sinus pauses >3 sec, may present as syncope Exercise intolerance Blood pressure dysregulation 	<ul style="list-style-type: none"> Surveillance annually with Echocardiogram, 24-72 hr Holter or implantable cardiac monitor Consider cardiopulmonary exercise testing Low threshold for pacemaker placement

	Prevalence (%) N=72, age=16 yr
Life threatening >=3sec systolic pauses or >2.5 seconds + syncope	22% (16/72)
Pacemaker placement	12/16
Symptoms	11/16
Syncope	8
Dizziness	3
Chest Pain	2
Tingling left arm	1
Palpitations	1

European Journal of Pediatrics (2020) 179:821–825
<https://doi.org/10.1007/s00431-019-03568-5>

ORIGINAL ARTICLE

Life-threatening cardiac arrhythmias in congenital central hypoventilation syndrome

Eric Laifman¹ · Thomas G. Keens^{1,2} · Yaniv Bar-Cohen^{1,2} · Iris A. Perez^{1,2}



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Cardiac Management

Clinical findings	Management Approaches
<ul style="list-style-type: none">▪ Bradyarrhythmia▪ Sinus pauses >3 sec, may present as syncope▪ Exercise intolerance▪ Blood pressure dysregulation	<ul style="list-style-type: none">▪ Surveillance annually with Echocardiogram, 24-72 hr Holter or implantable cardiac monitor▪ Consider cardiopulmonary exercise testing▪ Low threshold for pacemaker placement

- Annual Echo, Holter
- Get pacemaker if needed prior to transition
- Adjust/establish appropriate support during exercise

- Understand that high blood pressure is COMMON in adults – but may be masking pulmonary hypertension or part of CCHS dysautonomia.



GI Management

Clinical findings	Management Approaches
<ul style="list-style-type: none"> ▪ Hirschsprung Disease ▪ Gastroesophageal reflux disease ▪ Esophageal and Intestinal dysmotility ▪ Chronic constipation ▪ Dysphagia or oral aversion ▪ Need for gastrostomy tube 	<ul style="list-style-type: none"> ▪ Evaluation at diagnosis ▪ Further treatment and surveillance as needed based on symptoms and <u>findings</u>; low threshold for referral with refractory constipation ▪ For those with HSCR, bigger impact on QOL than the vent management

Prevalence (%)		
	n=72 ¹	n=196 ²
HSCR	31	16
GER	11	18
Constipation	17	23
G-tube	26	
Achalasia	1	
Esophageal/ other dysmotility	1	12
Chronic diarrhea		19
Absent gag		11
GENOTYPE MATTERS – Higher PARM and NPARM worse		

GI Management

Clinical findings	Management Approaches
<ul style="list-style-type: none">▪ Hirschsprung Disease▪ Gastroesophageal reflux disease▪ Esophageal and Intestinal dysmotility▪ Chronic constipation▪ Dysphagia or oral aversion▪ Need for gastrostomy tube	<ul style="list-style-type: none">▪ Evaluation at diagnosis▪ Further treatment and surveillance as needed based on symptoms and <u>findings</u>; low threshold for referral with refractory constipation▪ For those with HSCR, bigger impact on QOL than the vent management

- Determine motility issues prior to transition
- Understand that chronic abdominal discomfort/constipation/bloating is COMMON in adults – but may be masking underlying dysmotility or undiagnosed HSCR.



Perioperative Management

Clinical findings

Management Approaches

- Cardiorespiratory arrest with anesthesia, sedation, pain medications, recreational alcohol, or illicit drug use

- Pre-anesthesia evaluations
- Ambulatory procedures: (dental) – nitrous oxide (alone) plus local, continuous oximetry, BiPAP ready – otherwise do not do ambulatory!
- Provide education to operative room staff, even if at a tertiary center.
- Critical need for objective monitoring with any anesthesia, sedation, pain medications. Consider ICU

- Get as many procedures as you need done prior to transition!
- Say NO to ambulatory surgical centers – anything under anesthesia needs to be done in the OR

Pediatric Pulmonology 41:283–285 (2006)



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Alcohol/Drugs

Clinical findings

- Cardiorespiratory arrest with anesthesia, sedation, pain medications, recreational alcohol, or illicit drug use

Management Approaches

- Pre-anesthesia evaluations
- Ambulatory procedures: (dental) – nitrous oxide (alone) plus local, continuous oximetry, BiPAP ready – otherwise do not do ambulatory!
- Provide education to operative room staff, even if at a tertiary center.
- Critical need for objective monitoring with any anesthesia, sedation, pain medications. Consider ICU
- Pre-emptive counseling starting in early adolescence of alcohol/drug use and association with respiratory depression

Alcohol Use in Congenital Central Hypoventilation Syndrome

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Julienne R. Jacobson, MD,² and Thomas G. Keens, MD¹



Pediatric Pulmonology 41:283–285 (2006)



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Endocrine/Reproductive Management

Clinical findings

- Glucose dysregulation
- Isolated reports of other hormonal imbalances (GH, Thyroid)
- Body temperature dysregulation, low basal temperature
- Abnormal diaphoresis

Management Approaches

- Evaluation as needed, particularly in setting of unexplained syncope or seizures
- Do not rely on fever as symptom
- Symptomatic support for hyper- or hypothermia
- Discuss reproductive health, family planning given autosomal dominant transmission. Vigilant ventilatory monitoring during pregnancy and labor/delivery.



Endocrine/Reproductive Management

Clinical findings	Management Approaches
<ul style="list-style-type: none">▪ Glucose dysregulation▪ Isolated reports of other hormonal imbalances (GH, Thyroid)▪ Body temperature dysregulation, low basal temperature▪ Abnormal diaphoresis	<ul style="list-style-type: none">▪ Evaluation as needed, particularly in setting of unexplained syncope or seizures▪ Do not rely on fever as symptom▪ Symptomatic support for hyper- or hypothermia▪ Discuss reproductive health, family planning given autosomal dominant transmission. Vigilant ventilatory monitoring during pregnancy and labor/delivery.

- Obesity, diabetes common in young adults
- Sexual activity, pregnancy planning/prevention



Neurologic Management

Clinical findings	Management Approaches
<ul style="list-style-type: none">▪ Seizures▪ Syncope	<ul style="list-style-type: none">▪ Evaluation as needed to identify triggers, commonly cardiac, suboptimal ventilatory support, or endocrinopathy▪ Treatment for epilepsy per neurology

- Headaches, fatigue = adulting in general!
- But also may be related to CCHS



Final Thoughts

- Transition to adult care starts in the tween-teen years
 - Knowledge of CCHS
 - Ability to self-administer medical care
 - Reliable communication
- Unrealistic to continue going to a pediatric specific center – we need to develop training and pipeline for adult CCHS experts.

Thank you! – and next Dr. Orr!



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