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CHARGE OVER THE LIFESPAN: DEVELOPMENTAL, MEDICAL, BEHAVIORAL, AND TRANSITION ISSUES

C.H.A.R.G.E.

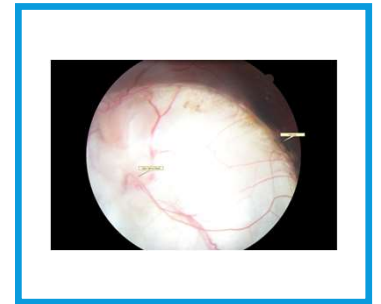
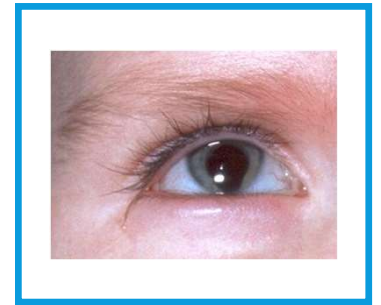


Let's begin with the acronym.



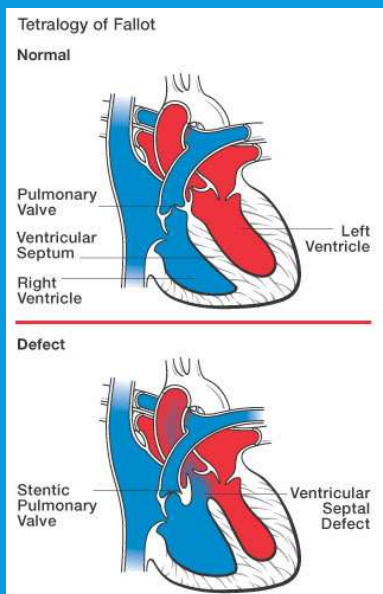
C - COLOBOMA

- An incomplete congenital development of various structures of the eye(s) or visual system, resulting in varying degrees of vision loss and/or interference.
- 80 - 90% of people with CHARGE syndrome may have a coloboma.
- Cannot be corrected with surgery.



H – HEART MALFORMATIONS

One heart defect
found in CHARGE

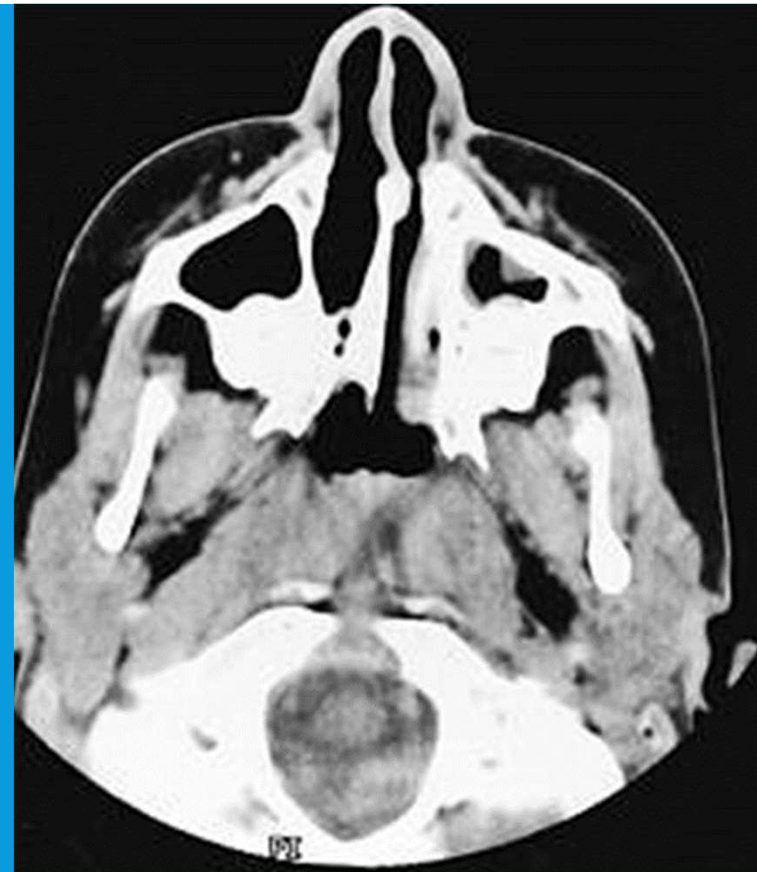


- Some defects are minor, some are complex.
- Most defects require surgery to repair.
- Occurs in about 65-85% of individuals with CHARGE.

This boy with CHARGE died from pulmonary atresia, with collateral arteries.

A – ATRESIA OF THE CHOANAE

- Choanae are the passages at the back of the nose leading to the throat that allow for breathing.
- Complete blockages are called “atresia”
- Narrowing of the passages is called “stenosis”
- Choanal atresia occurs in about half of the individuals with CHARGE.
- Often requires surgery to correct.
- 50-60% incidence



R – RESTRICTION IN GROWTH AND DEVELOPMENT

- Many individuals will have delays developmentally or be smaller than average in their growth (70% incidence).
- There is a wide range of ability in children from profoundly intellectually disabled to university graduates.

G - GENITOURINARY ABNORMALITIES

- Micropenis and undescended testes very common in males, under-developed labia can occur in females. (50% incidence)
- Delays in puberty occur in both sexes.
- Many different types of urinary tract anomalies associated with CHARGE (30-40%).

E – EAR MALFORMATIONS/HEARING LOSS

- Malformed ears very common in CHARGE syndrome (floppy, small lobes).
- Varying levels of hearing loss – can be mild, moderate to profound.
- Hearing loss sometimes difficult to evaluate.

CHARGE Outer Ears



BLAKE DIAGNOSTIC CRITERIA: THE FOUR C'S

Coloboma

Choanal Atresia

Cranial Nerve Dysfunction

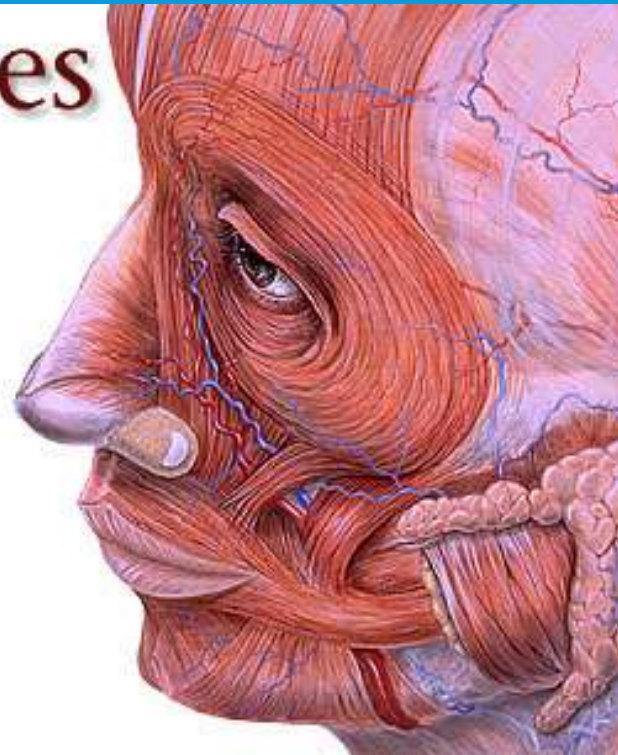
Characteristic CHARGE Ear



Dr. Kim Blake

Cranial Nerves

- I Olfactory
- II Optic
- III Oculomotor
- IV Trochlear
- V Trigeminal
- VI Abducens
- VII Facial
- VIII Vestibulocochlear
- IX Glossopharyngeal
- X Vagus
- XI Accessory
- XII Hypoglossal



Blake, K.D., Hartshorne, T. S., Lawand, C., Dailor, A. N., & Thelin, J. W. (2008). Cranial nerve manifestations in CHARGE syndrome. *American Journal of Medical Genetics*, 146A, 585-592.

SHOULD WE ADD TEMPORAL BONE ANOMALIES?

Balance in CHARGE

Average age of walking is three years old.

Range is 18 months to about 12.

Age of walking has been associated with:

- Adaptive behavior
- Challenging behavior
- Executive dysfunction
- Number of psychotropic medications
- Communication skills
- Sleep disturbances

MANY WITH CHARGE DON'T HAVE THIS
REFLEX.

Vestibulo-Ocular Reflex



www.SolveLearningDisabilities.com

How does this affect their functioning in the world?

SOLUTION:
Stabilize
the head!



SENSORY DEFICITS IN CHARGE

MULTISENSORY IMPAIRMENT

- Hearing – sensorineural hearing loss
- Vision – coloboma
- Smell – anosmia
- Taste – prefer strong tastes
- Tactile – defensiveness
- Vestibular – balance issues
- Proprioceptive – awkwardness



MAJOR MEDICAL MANAGEMENT ISSUES

- Breathing
- Heart defects
- Swallowing and Reflux
- Cleft lip and/or cleft palate
- Eating
- Motility and Gas
- Ear infections
- Puberty: Hormone replacement therapy
- Bone health: Osteoporosis and Spine (scoliosis)
- Kidney reflux
- Pain



Some see things; and say 'Why?' But I dream things that never were; and I say, "Why not?" —George Bernard Shaw

You are invited to Jacob Hartshorne's Future

*Please come and help unlock the doors into my exciting future, by
participating in my Person-Centered Plan!*

When: February 15, 2007

Time: 3:00-5:30 p.m.

Where: St. John's Episcopal Church,
Mt. Pleasant (406 W. Maple, corner of
Maple and Washington).

What is a Person-Centered Plan? A team
approach to help plan my future.

Why are you invited? You know me in a way
that no one else does. You are important to me!

What to bring? An open, creative mind and a
loving heart. Also, your appetite—we'll eat!

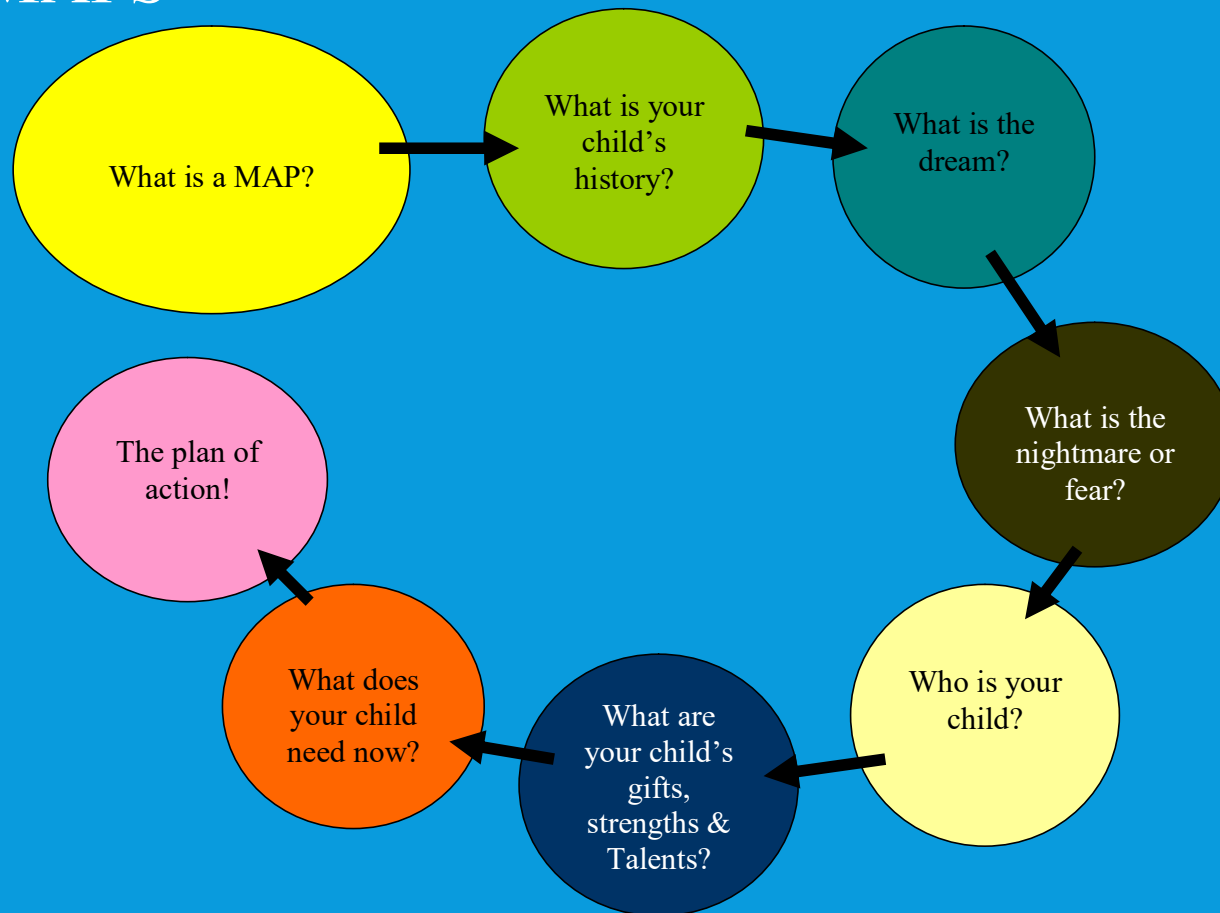


RSVP to DB Central, Michigan Services for
Children and Youth who are Deafblind (989) 774-2725
Or reply via email: nancy.hartshorne@cmich.edu



Together we can make my future bright!

PLANNING MAPS



HISTORY



- What people and events have shaped this person's life?
- What have been the highlights?
- How have others present been a part of this history?

DREAMS

- Short and long term future
- May be open ended
- Dreams do not have to be “realistic”
- No dream is too big or too small
- Dreams give hope and a possible direction

FEARS / NIGHTMARES

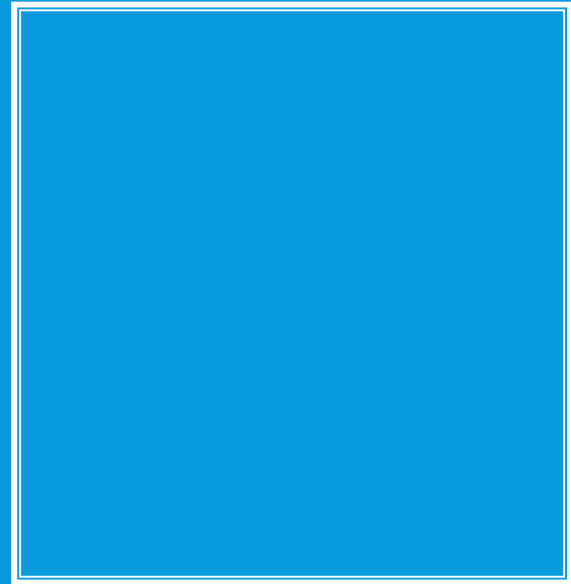
- Barriers to realizing dreams
- May be uncomfortable or revealing
- May be emotionally charged
- How can fears and nightmares be avoided?

WHO IS ...?

- Strengths
- Skills
- Likes and dislikes
- Personal qualities
- Favorite activities
- Friends
- Are not always totally positive, but need to be respectful

NEEDS

- Hopes, strengths, interests
- Activities, opportunities, supports
- The focus is positive
- Use brainstorming



ACTION PLAN

- Move toward the dream while avoiding the nightmares
- What would a perfect day be like for this person?
- Who is willing and able to support?
- What, Who, and By When
- Make sure there is a clear plan for follow-up
- Use available agency supports AND natural supports.

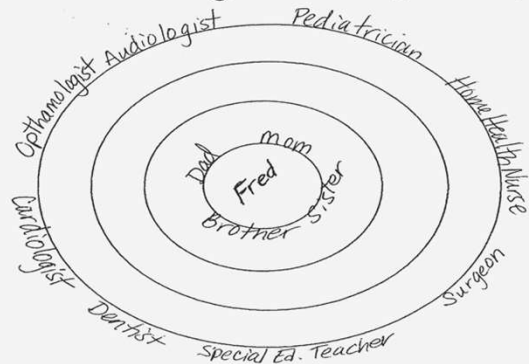


NO PLAN IS PERFECT

- Plan early – Kindergarten is not too soon
- Transition services are essential
 - Survival communication (signs, pictures, etc.)
 - Job skills
 - Diploma vs certificate
 - Agency identification
- Networking
 - Finding the network
 - Information, resources, connection
 - Do not go it alone
- Advocate
 - Make it their problem, not just yours
- Develop skills and find resources
- Keep person-centered planning

GOALS

ed has a severe disability. He has attended a segregated setting for eight years



CIRCLES OF FRIENDS

Who supports you?

1. **Circle of Intimacy:** people most intimate in your life – you can't imagine living without them
2. **Circle of Friendship:** very close friends with whom you choose to spend time
3. **Circle of Participation:** people, teams, organizations, and networks with whom you are involved (colleagues at work, members of your church, your softball team)
4. **Circle of Exchange:** people you pay to provides services in your life (doctors, dentists, tax accountants, mechanics, teachers, store clerks)

From:

Perske, R. (1989) *Circles of friends*. Nashville: Abingdon Press.
 Falvey, M., Forest, M., Pearpoint, J., Rosenberg, R. (1994). Building Connections. In J. Thousand, R. Villa, & A. Nevin (Eds) *Creativity and Collaborative Learning*. Baltimore: Paul H. Brookes.

Being Present And
Participating In
Community Life

Gaining And
Maintaining
Satisfying
Relationships

Expressing
Preferences And
Making Choices

Continuing To
Develop Personal
Competencies

Fulfilling Respected
Roles And Living
With Dignity

BARRIERS AND SUPPORTS

- Sensory – deafblind intervener
- Medical – management team
- Communication – choose an approach early
- Services – identify what is available, and insist on what else is needed
- Funding – keep asking
- Planning – Person Centered Planning

QUALITY OF LIFE

- Love
- Friendship
- Work
- Health, sleep, anxiety
- Housing, transportation, service availability

BIRTH AND THE NICU

- Shock
- Will my baby live
- What is wrong with my baby
- Can I keep my baby alive

NEEDS - WHAT DO PARENTS NEED TO KNOW?

- About the condition and how to deal with it
- About how to find support
- About finding treatment and intervention
- About everyday care
- About finding equipment
- About the future and how to plan for it
- About how to talk with others about the experience
- About how to get organized around situations
- About how this affects the family as a whole

Adler, Salanterä, Leino-Kilpi, & Grädel (2015)

HOW DO YOU FIND OUT?

- Research it
- Ask professionals
- Ask other parents
- Ask the network

"In every community there is generally a network of people who get disability, who support one another, and who are knowledgeable regarding services and service providers."



THE FIRST FOUR YEARS: STAYING ALIVE

- Heart surgery
- Choanal Atresia repair/cleft repair
- Aspiration pneumonia
 - Tracheostomy
 - Gastrostomy
- Ear infections
- Multiple surgeries

NEEDS

- Medical training
- Equipment
- Identifying services
- Balancing care and work
- Sleep
- Emotional self-care
- Access to quality medical care
- Network and social support
- Adjusting to the new normal

Major decisions regarding lifestyle, support of family members, work-home balance, and making major medical decisions, anticipating school placement decisions, and selecting a communication system for your child.

COMMUNICATION SYSTEM

- American Sign Language
- Signing Exact English
- Oral/Aural
- Picture Exchange
- Whole Language

THE START OF SCHOOL

- Your child does not belong
- We do not provide those services
- Listen to the experts
- Lower expectations

NEEDS

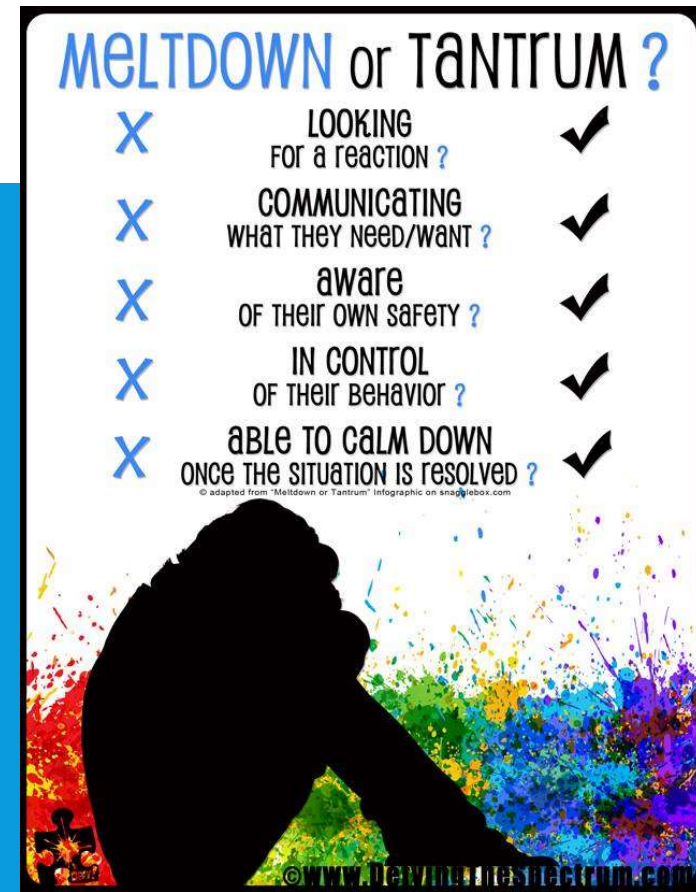
- An ally
- A degree in special education
- Understand IEP's and the law
- How to fight without blowing up bridges
- Make placement decisions
 - Inclusion
 - Mainstream
 - Regular classroom
 - Restricted classroom
- Communication system
- Request services
- Circle of friends

“It is the parents’ responsibility to make educational choices for their child. It is the school’s responsibility to make them work.”

BEHAVIOR

- Behavioral challenges are not inevitable
- No child with CHARGE should be without access to
 - Sensory breaks
 - Calendar system
 - Social stories
 - Circle of Friends
- Decisions
 - Diagnosis
 - Medication

SENSORY BREAKS





CALENDAR SYSTEMS



Anxiety due to a lack of predictability
Utilize an informed calendar intervention
Increase self-regulation and predictability
Reduce anxiety and anxiety related behaviors

Get Ready For Bed



SOCIAL STORIES



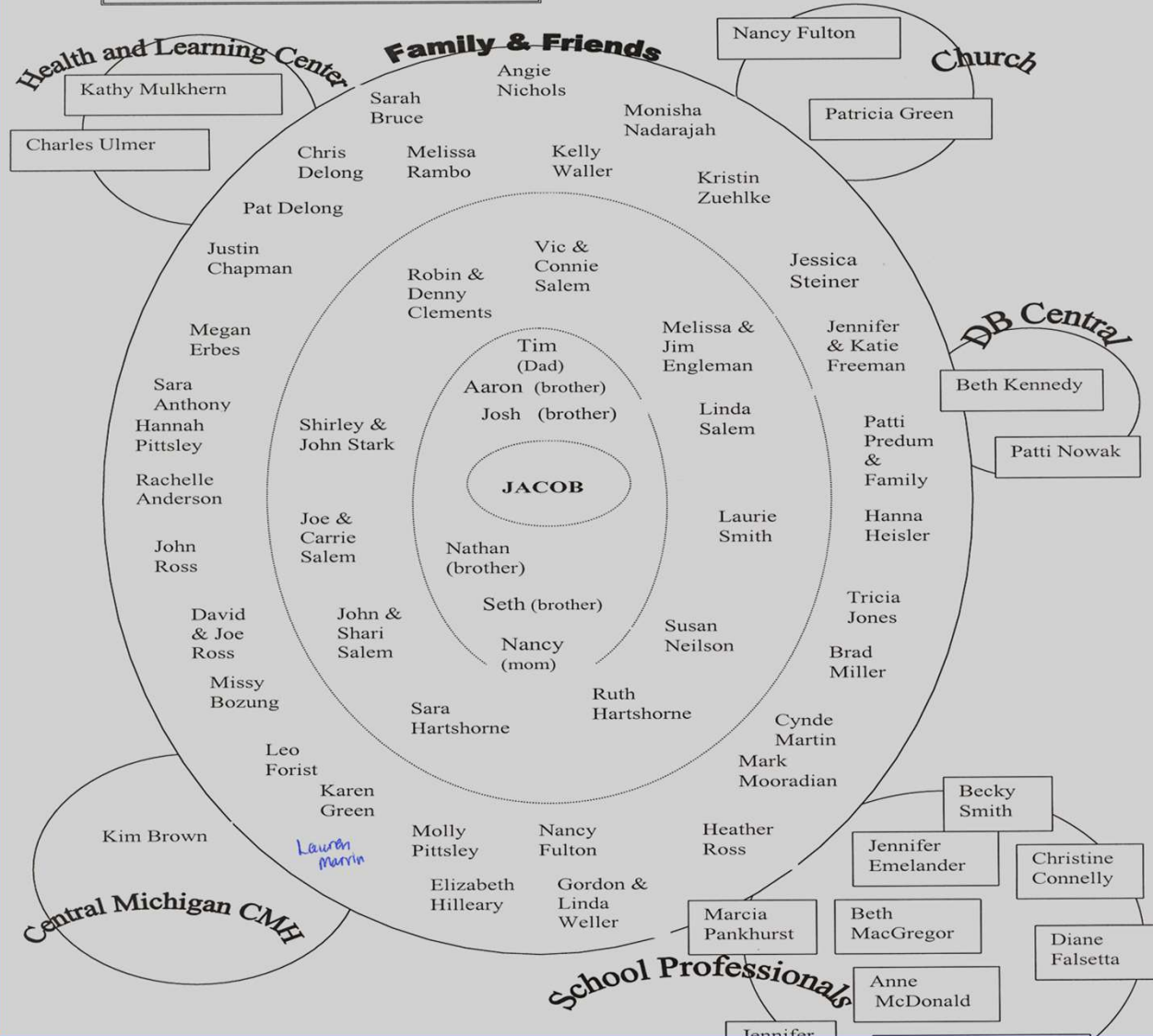
Short, simple, to the point, easy to make, personalized
Conveys social situation and explains how someone would react
or what to do

Repetition makes the message stronger

Can help teach a lesson visually

Potty training, sharing, handling anger, change in routine, etc.

RELATIONSHIPS MAP



MIDDLE SCHOOL

- Multiple teachers
- Transition time between classes - LOCKERS
- Greater emphasis on teacher control and discipline
- Less personal and positive student-teacher relationships
- Increased use of whole-group instruction
- Peer struggles

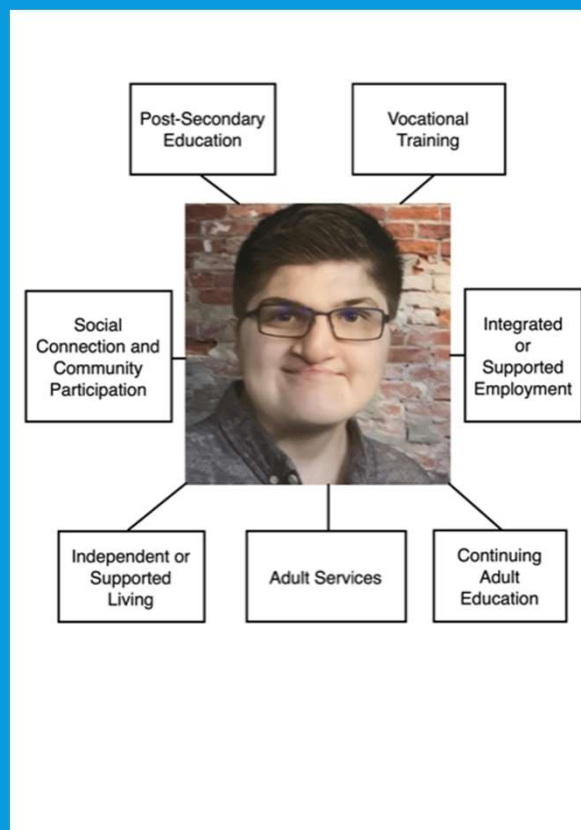
NEEDS

- The principal and school secretary must be on your side
- Educating teachers
- Parallel curriculum
- Circle of friends
- Decision – how much time in special education

HIGH SCHOOL

- May be in a more restricted placement
- Less access to peer group (but they may still care)
- Transition issues predominate

AN ADOLESCENT WITH CHARGE IS CONFRONTED BY MANY ISSUES.



TIME TO LEAVE SCHOOL

- With what?
- To what?
- How?

TRANSITION SERVICES

- We started planning way ahead. Met with Lacey's school team and community bridges regularly to plan. I talked with other families. I found a program in our small town that I was excited about, farm based. Thought this would be great for Lacey as she loves getting her hands dirty and messy. Loves gardening! I met with the director, saw the program and Lacey went with her school team and spent some hours there. She did well there. 2 weeks before Lacey was going to graduate, I got an email, not even a personal phone call, saying Lacey was too medically involved for their program after they had already said yes. We had been planning with them for a year. I was so upset but something bad turned into something good and we found something better. A place where they were more accepting, and she was wanted.

ADULTHOOD

"I would just like to be able to live by myself, to have friends and a social life, and to have a job."

But this takes a lot of planning, and transition services should be addressing all of the following.

CHALLENGES IDENTIFIED BY THE EXPERTS

Accessing services

- Not knowing what is out there or possible
- Services denied
 - Does not qualify
 - Medicaid will not cover

Day programs

- Will they do medical care like g-tube
- Communication needs
- Behavior
- Most seem to be for higher functioning or for seniors
- Funding
- Will they follow the Person-Centered Plan?

MORE CHALLENGES IDENTIFIED BY PARENTS

Adult medical
care

Funding for
hearing aids

Transportation

Employment

Finding
activities – red
tape

Security -
trusts

HOUSING

1

Live with parents
in parents' home

2

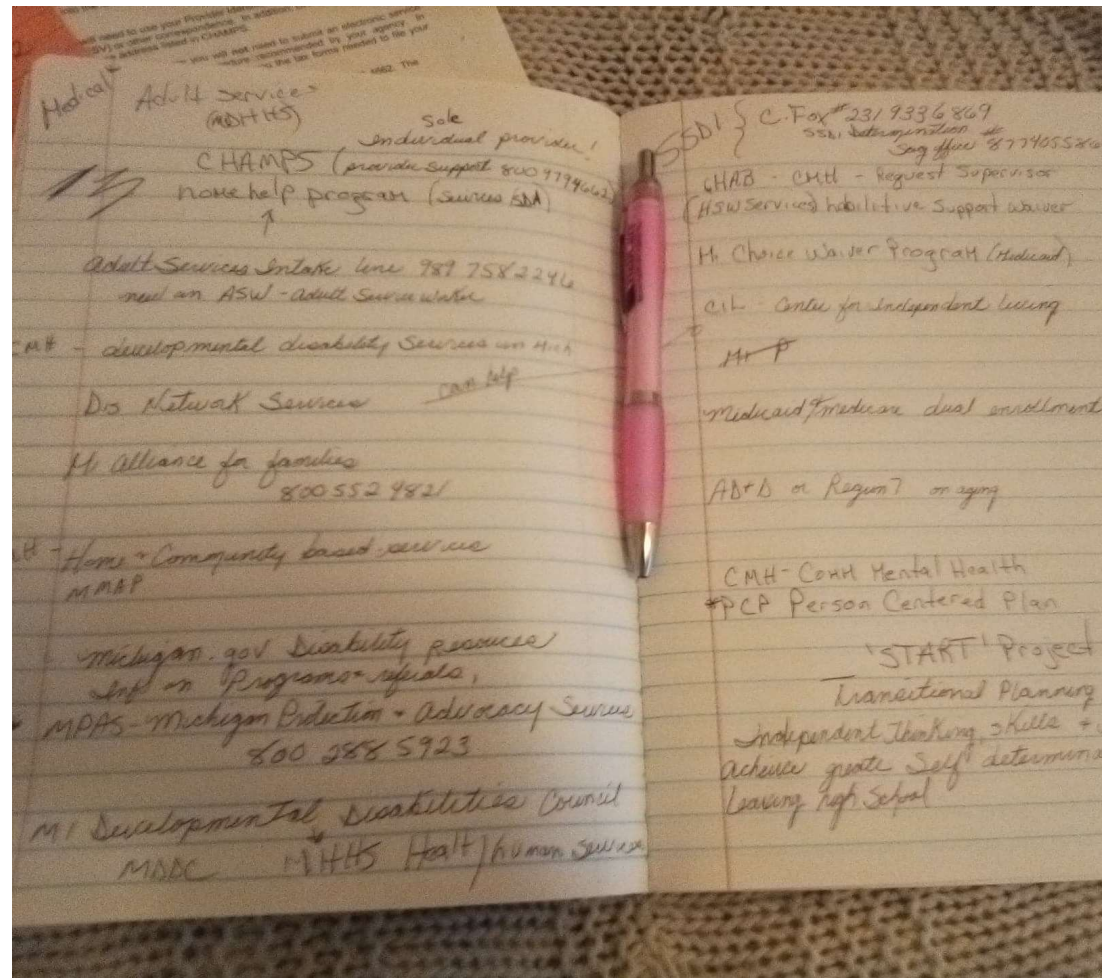
Live in group
home or
institution

3

Live
independently
with supports

SERVICES

I am so overwhelmed with services and programs and resources with research I've been doing I just can't figure out where we fit. I was up all night. I'm just so all over the place here with trying to learn where to start on adult services and just which agency. I've got social workers flustered on helping guide me asking their supervisors for direction. I'm just getting bounced around. This mom is about spent sigh.



GUIDING PHILOSOPHY

The best (effective & efficient) way to meet people's needs is by supporting them in the life they want!

But this takes PLANNING.

LIFE CAN BE GOOD



Thanks to my Lab

CHARGE Syndrome Research Lab
At Central Michigan University

