

GENETIC COUNSELING AS A COMPONENT OF THE NEWBORN SCREENING SYSTEM

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Assuring clinical genetic services for newborns identified through US newborn screening programs.

Kaye, CI, et al. *Genet Med.* 2007; 9(8): 518 - 527.

Newborn screening follow-up: approved guideline.

Wayne, PA. CLSI. 2006; 26 (18).

Genetic counseling and neonatal screening for cystic fibrosis: an assessment of the communication process.

Ciske DJ, et al. *Pediatrics.* 2001; 107: 699-705.

US newborn screening system guidelines II: follow-up of children, diagnosis, management, and evaluation. Statement of the Council of Regional Networks for Genetic Services (CORN).

Pass KA, et al. *J Pediatr.* 2000; 137(4 Suppl): S1-46.

OBJECTIVES

○ Genetic counseling

- What is it?
- What is the goal of NBS and of genetic counseling within the NBS system?
- Who provides genetic counseling?

○ Oklahoma example

○ Considerations for state NBS systems

NSGC definition of genetic counseling

(Resta, et al, 2006)

Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates the following:

- *Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.*
- *Education about inheritance, testing, management, prevention, resources, and research.*
- *Counseling to promote informed choices and adaptation to the risk or condition.*

What are the goals of NBS and genetic counseling within that system?

- Goal of NBS: *“The most important and widely accepted goal of newborn screening is to improve health outcomes in the screened population of newborns.”*

Moyer VA, et al., on behalf of the United States Preventive Services Task Force. "Expanding Newborn Screening: Process, Policy, and Priorities," *Hastings Center Report* 38, no. 3 (2008):32-39.

- Goal of genetic counseling within NBS system:
What is it?????

Who provides genetic counseling?

- Genetic counselors
- Pediatricians
- Newborn screening coordinators
- Nurses
- Single gene educators
- Medical geneticists

OK genetic counseling sites



Genetic counseling in OK NBS follow-up program

- Genetic counseling (by a genetic counselor) was provided, as needed, as part of the LTFU metabolic program (PKU, galactosemia, MCADD), but no contract for services existed.
- NBS for cystic fibrosis began in 2005. Since that time, 98% of CF screen positive cases with one and two mutation(s) have received genetic counseling.
- Contracted genetic counseling services for hemoglobinopathies was incorporated the following year.

CF screen positive protocol

- Elevated IRT leads to DNA (44 mutations at OSDH); if mutation identified, then.....
- STFU notifies pediatrician and tertiary center.....
- Pediatrician refers infant for sweat test
 - Mondays (OKC) and Fridays (Tulsa) are dedicated days
 - Sweat lab contacts GC
- GC meets with family after the sweat while results are pending and collects a family history.....
- GC receives call from the sweat lab with results and reports them to the family
- Infants diagnosed with CF are referred to the CF Center
- Pediatrician and families receive a counseling letter

Cystic Fibrosis Newborn Screening - Genetic Counseling Report Form

Reporting requires this form be completed and submitted within 24 hours and a copy of the genetic counseling follow-up letter be submit within 14 days to the Newborn Screening Program, fax (405) 271-4892 address 1000 NE 10th Street, Oklahoma City, OK 73117-1299. Information call (405) 271-6617.

Newborn's Name: _____ LAB# _____ DOB: ____/____/____

Appointment date: ____/____/____ Date of Counseling Session: ____/____/____

Time of Counseling Session: _____ am ____ pm Name of Counselor: _____
Print Last Name, First

of Family Members Present for Session _____ Family Members Present for Session (check all that apply): Mother Father Grandparent Other

Date of Sweat Test: ____/____/____ Time _____: ____ am ____ pm

Sweat testing site: OU Medical Center, OKC Saint Francis, Tulsa Other: _____

Results: _____ > 60 mmol/L _____ 30 to 60 mmol/L _____ < 30 mmol/L _____ QNS Repeat Sweat ____/____/____

DNA Results: _____ 0 mutations _____ 1 mutation _____ 2 mutations REFLEX TESTING ORDERED: YES NO

Family Carrier Testing: _____ Yes _____ No _____ Undecided If yes, check who was tested: _____ Mom _____ Dad (other) _____

Check all that apply:
 No show for sweat testing
 No show for genetic counseling
 Family refused counseling
 Session not performed on the same day as the sweat test

For sections I & II, check all that was completed during the face-to-face counseling session:

I. Information obtained and client assessments:

- Reviewed family's questions and reason for referral
- Assessed knowledge base, perception of disease status/risk, perceived notion about causation, need for interpreter
- Assessed family history (1st and 2nd degree relatives to consult, and further removed as appropriate) for information pertinent to CF including but not limited to perinatal losses (meconium ileus) and childhood deaths, chronic lung disease, pancreatic insufficiency, infertility, ethnicity, consanguinity
- Psychosocial assessment (i.e., personal, social, religious, ethnocultural issues, family functioning)
- Assessed ethical concerns, such as confidentiality and non-paternity

II. Information provided and discussed:

- Results of newborn screen and sweat test
- Relevant tests for patient and/or other relatives (sweat testing, CFTR mutation analysis, DNA linkage studies)
- Sensitivity/specificity of tests
- Addressed psychosocial issues
- Provided resources as appropriate

III. To improve service delivery, please provide any additional comments and constructive feedback from the family:

Select session type, and check all components covered during the face-to-face counseling session:

No mutations, negative sweat test (unaffected) the session shall include:

- Limitations of testing
- Other causes of elevated IRT (possible carrier of mutation not on panel)
- Encouraged regular check-ups with pediatrician

One mutation present, negative sweat test (Carrier) the session shall include:

- Explanation of inheritance pattern including possibility both parents are carriers
- Explanation of risk to other family members including plan for relaying information
- Offered parental carrier testing
- Carrier status is not associated with health complications
- Provided carrier brochure

Two mutations present, negative sweat test (affected) the session shall include:

- Natural history including variability of clinical phenotype
- Explanation of inheritance pattern including recurrence risk
- Explanation of risk to other family members and plan for relaying information
- Future reproductive options
- Further testing options (repeat DNA analysis and parental carrier testing)
- Discussed genotype-phenotype correlations if known
- Recommended additional genetic counseling if indicated
- Addressed psychosocial issues including anticipatory guidance and reaction to diagnosis
- Referred to CF specialty clinic and provided resources

Zero to One mutation, positive sweat test (affected) the session will include:

- Natural history
- Explanation of inheritance pattern to include recurrence risk
- Explanation of risk to other family members including plan for relaying information
- Future reproductive options
- Further testing options (expanded mutation panel testing of infant, parental carrier testing and linkage analysis)
- Recommended additional genetic counseling as indicated
- Addressed psychosocial issues including anticipatory guidance and reaction to diagnosis
- Referred to CF Center and provided resources

Two mutations, positive sweat test (affected) the session shall include:

- Natural history
- Explanation of inheritance pattern to include recurrence risk
- Explanation of risk to other family members including plan for relaying information
- Future reproductive options
- Further testing options to include additional mutation testing of infant if indicated and parental carrier testing
- Discussed genotype-phenotype correlations if known
- Recommended additional genetic counseling if indicated
- Addressed psychosocial issues including anticipatory guidance and reaction to diagnosis
- Referred to CF specialty clinic and provided resources

Zero to two mutations, with borderline sweat test (status unknown) the session shall include:

- Natural history including variability of clinical phenotype
- Limitations of test
- Explanation of inheritance pattern to include recurrence risk if applicable
- Explanation of risk to other family members including plan for relaying information
- Future reproductive options
- Further testing options (expanded mutation panel testing of infant, linkage analysis and parental carrier testing when clinically appropriate)
- Discussed genotype-phenotype correlations if known
- Recommended repeat genetic counseling (infants expected to have a repeat sweat test in 4 weeks)
- Referred to CF specialty clinic if two mutations were present
- Addressed psychosocial issues including anticipatory guidance and reaction to uncertainty of diagnosis
- Provided resources

Comments _____

If components of the session were not covered (I, II & III), explain why e.g., parent not receptive to information, parents not attentive, infant disrupted session etc.:

Dear Parents:

When your baby was born, the hospital drew a newborn screening test to determine your baby's hemoglobin type. The results of this blood test indicate that your baby probably has **Sickle Cell Trait**. Sickle Cell Trait will not make your baby sick. Enclosed is a pamphlet about Sickle Cell Trait.

At this time, genetic counseling is needed. To schedule a **FREE** genetic counseling appointment call **(405) 271-8685**.

Factors promoting genetic counseling in OK NBS follow-up

- NBS program and specialty service providers have good working relationships
- Small state; two major cities
- Availability of trained genetic counselors
 - Genetic counseling training program
- CF center, Sickle cell center, metabolic program, and a LTFU program
- Reimbursement for genetic counselors

What could we do better?

- ◎ Telemedicine
 - Local health departments are wired
- ◎ Continuous quality improvement
 - Evaluate factors influencing uptake
- ◎ Research
 - Risk communication study

Issues to consider

- Genetic counseling capacity and access in your state
- Goals for genetic counseling in your state
- Preferred model and timing of genetic counseling
- Genetic counselors knowledge about NBS
- Cost of genetic counseling services
- Genetic counselors serve on your state Genetics/NBS Advisory Council

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<http://www.nsgc.org>

Regional Coordinating Center

<http://genes-r-us.uthscsa.edu/>