TRANSITION

EXAMPLE PACKET

Reno, NV
July 2003

Supportive Services-Important Numbers

SERVICE	NAME	NUMBER/ADDRESS
Service Coordinator		
Schools		
Employment		
Housing		
Transportation		
Family		
Doctors		
Dentist		
Speech Therapy		
Occupational Therapy		
Pharmacy		
Psychology		
State Dept. of Health		
County Dept. of Health		
Respite Care		
Waiver Coordinator		
Job and Family Services		
Insurance		
Others		

Clinician's Handbook of Preventive Services: Children/Adolescents: Screening

Chapter 8: Newborn Screening

Office of Disease Prevention and Health Promotion
Public Health Service
Peer Review Status: Externally Peer Reviewed

Newborn screening for congenital hypothyroidism and phenylketonuria (PKU) is required in virtually all states. Testing for galactosemia and hemoglobinopathies is required in a majority of the states. Other conditions for which some states require newborn screening are maple syrup urine disease, homocystinuria, biotinidase deficiency, tyrosinemia, congenital adrenal hyperplasia, cystic fibrosis, and toxoplasmosis. Screening for some conditions is voluntary in some states. See Table 8-1 for a state-by-state listing of newborn screening policies.

Hypothyroidism
Most children with congenital hypothyroidism who are not identified and treated promptly suffer the irreversible mental retardation and varying degrees of growth failure, deafness, and neurologic abnormalities comprising the syndrome of cretinism. Incidence is 1 per 3600 to 1 per 5000 live births. Infants who receive adequate treatment with thyroxine within the first weeks of life have normal or near-normal intellectual performance when tested at 4 to 7 years of age.

Phenylketonuria (PKU)
This autosomal recessive aminoacidopathy leads to severe, irreversible mental retardation (IQ below 50) when untreated during infancy. Incidence is 1 per 10,000 to 1 per 25,000 live births. With early screening, diagnosis, and optimal treatment with a dietary restriction of phenylalanine, most children are in the normal range of intelligence.

Galactosemia
This disease causes failure to thrive, vomiting, liver disease, cataracts, and irreversible mental retardation. Death often results from Escherichia coli septicemia. The incidence is 1 per 60,000 to 1 per 80,000 live births. Removal from the diet of galactose-containing foods, espe-cially milk, leads to a dramatic improvement in the patient, and all clinical features except mental retardation may improve or disappear.

Hemoglobinopathies
Sickle cell disease and other hemoglobinopathies, such as thalassemia and hemoglobin E, are most common in individuals of African, Mediterranean, Asian, Caribbean, and South and Central

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Stanton Segal

Galactosemia unsolved

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Abstract Classic galactosemia is an enigmatic disorder that presents the challenge of unraveling the basis of the long-term complications of mental disability, speech defects, ovarian failure and neurologic syndromes which occur despite a galactose-restricted diet. A complete understanding of the pathobiochemistry and molecular genetics, and evaluation of the present theories for the poor long-term outcome, continuous intoxication, critical metabolite depletion and in utero damage is needed in order to design new therapeutic strategies. Answering this urgent question of how to treat galactosemic patients mandates enhanced clinical and basic research efforts.

Key words Chronic intoxication · Complications · Endogenous galactose · Galactose metabolism

Abbreviations *GALT* galactose-1-phosphate uridyltransferase · *Gal-1-P* galactose-1-phosphate · *Glc-1-P* glucose-1-phosphate

Introduction

It is the aim of this paper to reflect on a disorder that has become an enigma: Galactose restriction has been the basis of therapy of congenital galactosemia due to galactose-1-phosphate uridyltransferase (GALT) deficiency since Mason and Turner [45] in 1935 described how removing galactose from the diet elminated the acute toxicity syndrome. The clinical picture of a galactosemic infant with severe inanition, cataracts, hepatomegaly and jaundice can be readily changed to a thriving child with regression of these symptoms and signs within a short time after a galactose-free diet is started. This marked improvement has caused many to feel that early diagnosis and institution of stringent dietary therapy would result in normal children. This appears not to be the case.

Despite early diagnosis and institution of a galactose-free diet a number of clinical observations have suggested that mental disability and speech impairment may occur [14, 41]. Ovarian failure was observed in afflicted females [36, 70], and reports appeared of an ataxia syndrome in some well-treated patients [16, 44]. A poor outcome has been highlighted in the recent retrospective survey of over 350 patients by Waggoner et al. [76], where developmental delay, speech impairment, ovarian dysfunction and growth retardation were found in a number of patients seemingly independent of the time that dietary restriction was begun. The high incidence of patients with such sequelae did not differ regardless of whether the patient had a normal neonatal history and treatment before the onset of symptoms or of whether the treatment ensued after symptoms were observed. It was apparent that the complications in later childhood seemed to be unrelated to the time treatment began within the first 2 months. These disturbing findings on the outcome of well-treated patients and those published by Schweitzer et al. [60], and reported by Naughton [47] and Bakker [2] form the enigma of galactosemia. The question is when galactose is eliminated from the diet on the 1st day of extrauterine life or from the mother during pregnancy, why are we not able to produce a normal individual?

Chatty Cathy
1234 America Street
Boom Town, OR 56789
(123) 444-5555

Jan. 1, 2000

Developmental Disabilities Board
456 Transportation Dr.
Boom Town, OR 56789

Attn: Mr. Blue

RE: Chatty Cathy
Claim Number 123-23-1234 *(usually social security number)*

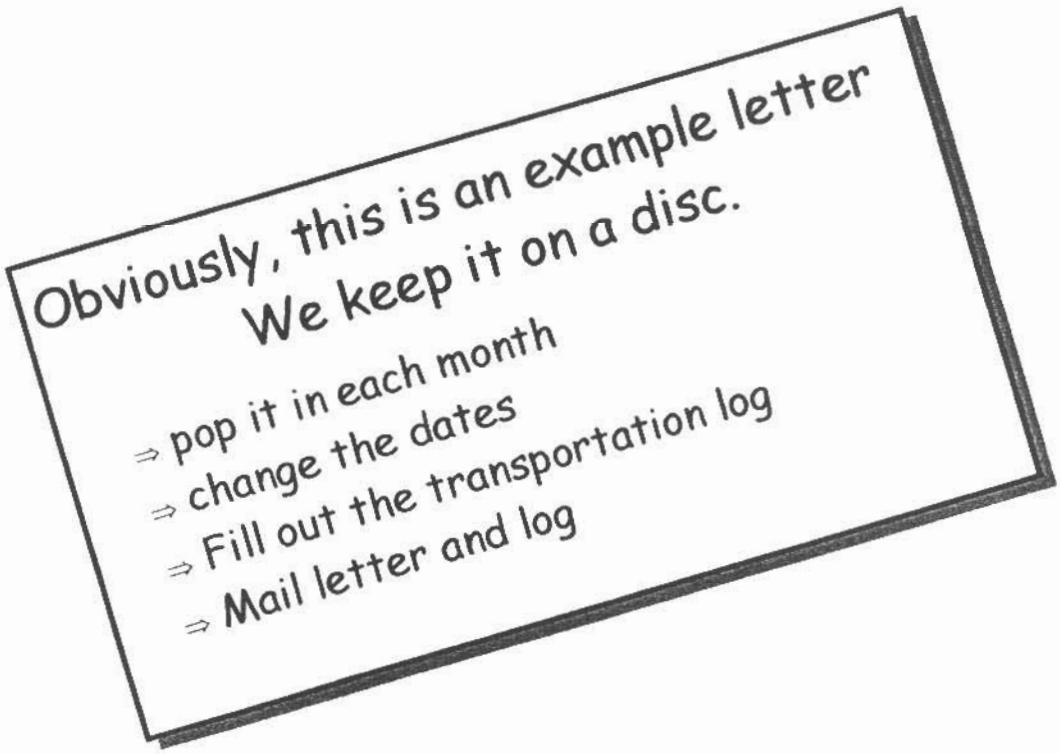
Dear Mr. Blue

Please find enclosed the transportation log for the month of January 2000 for Chatty Cathy, As per her ISA plan I am requesting reimbursement for transporting Cathy to and from work.

Again, thank you for your cooperation in this matter.

Sincerely,

Chatty Cathy



DELAWARE COUNTY BOARD OF DEVELOPMENTAL DISABILITIE

Scott Morgan
Superintendent

TRAVEL REIMBURSEMENT AUTHORIZATION

The Alpha
Henry Kho
Region

Employee Name: _____ Department: _____

DCBMRDD Drivers License No: _____ Expiration Date: _____

Period _____ In County? ☐ Yes ☐ No Page _____ of _____

DATE	DESCRIPTION/PURPOSE (Destination, Address, Remarks)	✓ CLIENT TRANS?	MILES	OTHE EXPEN

CERTIFICATION: I Certify That:

1. The statements made heron are true
2. Driving and other expenses were incurred on official business
3. This request is in compliance with DCBMRDD policy on the revese side of original authorization
4. I have vehicle liability insurance of $100,000/300,000/100,000 or combined single limits of 300,000.

TOTAL MILES _____

X 0.30

SIGNATURE _____ DATE _____ TOTAL EXPENSES _____

APPROVAL:

1. I concur with above expenses
2. I have checked license expiration

TOTAL REQUEST _____

APPROVAL _____ DATE _____

1000 Alpha Drive Delaware, OH 43015 (614)368-1751 (614)548-7133 Ext.1751

Chatty Cathy
1234 America Street
Boom Town, OR 56789
(123) 444-5555

Jan. 1, 2000

Social Security Office
123 Executive Dr.
Boom Town, OR 56789

Attn: Mr. White

RE: Chatty Cathy
Claim Number 123-23-1234 *(usually social security number)*

Dear Mr. White,

Please find enclosed the pay stubs for Chatty Cathy for the month of January 2000.

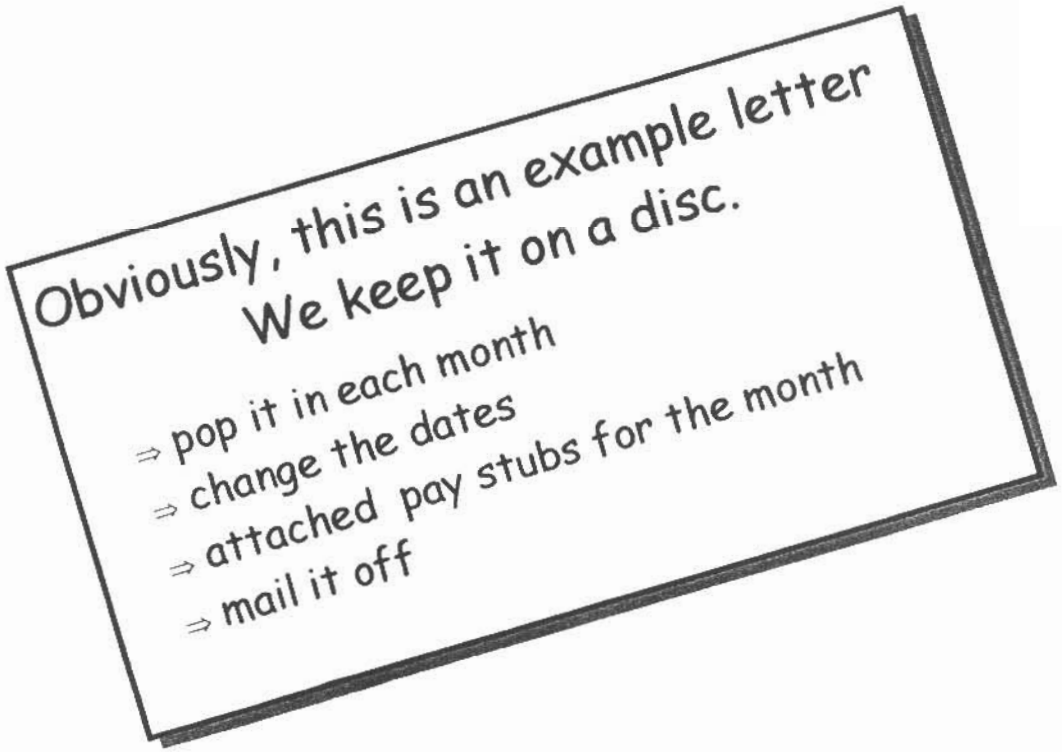
I trust as in the past the appropriate adjustments will be made.

If you have any questions please do not hesitate to call.

Thank you for your assistance in this matter.

Sincerely,

Chatty Cathy



Social Security

What You Need To Know When You Get SSI

Handy Booklet you can order

www.ssa.gov

Why You Should Read This Booklet

This booklet is for people who get Supplemental Security Income (SSI). The first part tells about your benefits and what you can expect from Social Security. The second part tells what changes you must report. The third part tells you how you can call Social Security toll-free to conduct your Social Security business. The fourth part tells how and when to report changes. The fifth part explains some special rules for people who get SSI disability. The sixth part of the booklet explains other things all SSI recipients should know, and the last part provides additional information about SSI eligibility.

Please take a few minutes now to read the booklet. Then put it in a safe place and look at it now and then. This will help remind you to report any changes to us and remind you of things you should know.

If you get Social Security retirement or survivors benefits, you also should read, *What You Need To Know When You Get Retirement Or Survivors Benefits* (Publication No. 05-10077). If you get Social Security disability benefits, you should read, *What You Need To Know When You Get Disability Benefits* (Publication No. 05-10153). You can get these booklets from Social Security.

Social Security's
Internet Website: *www.ssa.gov*
Toll-Free Number
1-800-772-1213
TTY Number
1-800-325-0778

Social Security Administration
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