Single Gene Disorders:
Cystic Fibrosis and Family History

Richard B. Parad, MD, MPH
Brigham and Women's Hosptial
Department of Newborn Medicine
Harvard Medical School, Boston, MA
and
New England Newborn Screening Program
University of Massachusetts Medical School

Family History for CF Risk Assessment

- As an autosomal recessive disorder, prior to the 1989 CFTR gene discovery, carrier status could only be identified by being the parent of an affected child, and other family members provided with a calculated risk, and dx only through sx or FHx.
- DNA diagnostics now allow direct carrier identification, but the test is imperfect, expensive and benefits from focus.
- Population screening for CF carrier status (national) and CF newborn screening (state based) are developments that change but do not preclude the importance of FHx.

Cystic Fibrosis
Clinical

- Lung Disease
  - chronic infection, inflammation and airway obstruction
- Gastrointestinal Disease
  - pancreatic insufficiency with fat malabsorption leading to malnutrion
- Salt Loss - with high sweat chloride
- Sterility in males
- Other
  - cirrhosis, diabetes

Estimated Incidence by Ethnic/Racial Background

- Caucasian 1/3,000
- Hispanic 1/6,000
- African-American 1/10,000
- Asian 1/30,000

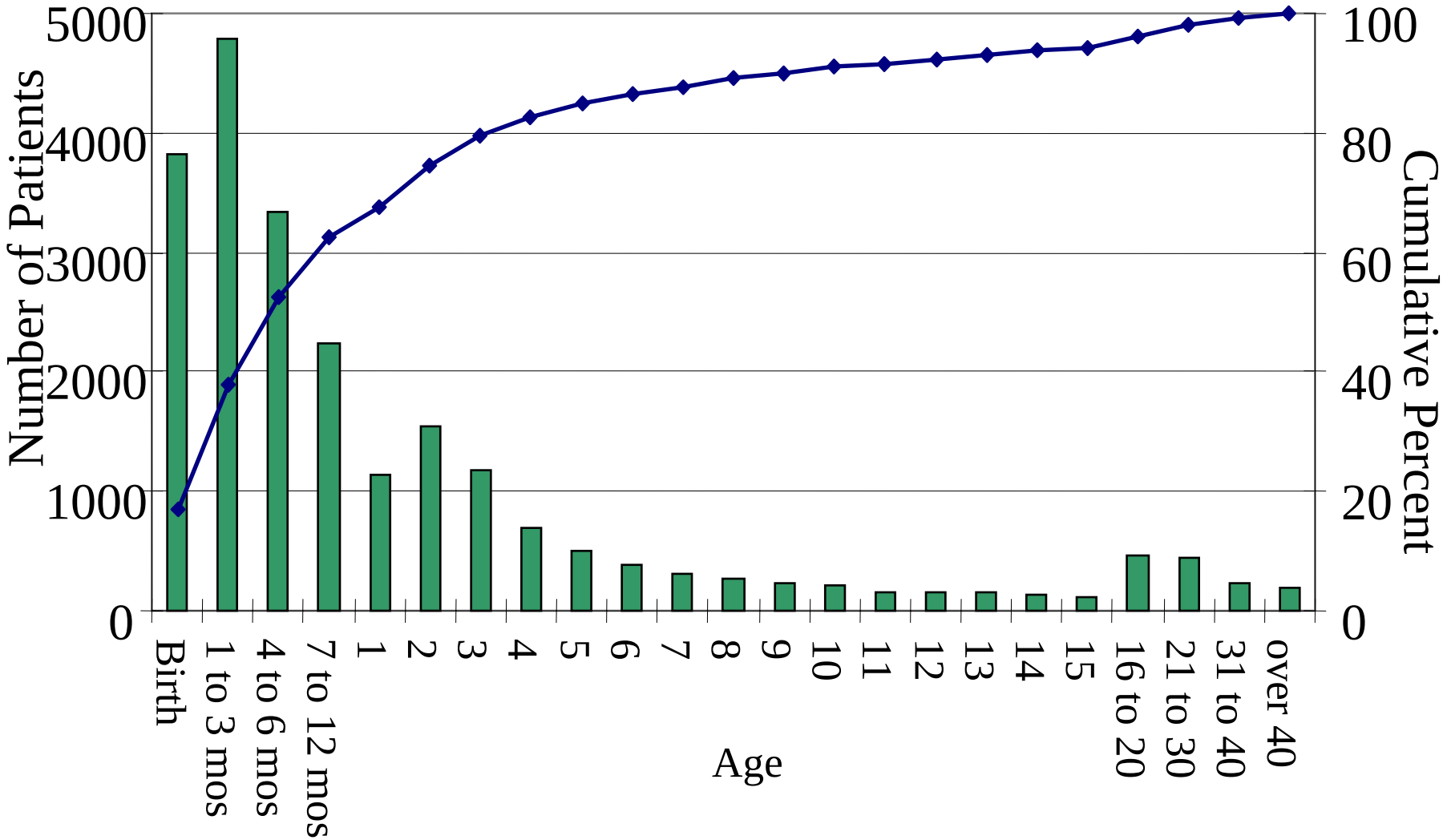
Symptoms in 864 Newly Diagnosed Patients With CF

RESPIRATORY	**52.1%**
Failure to thrive/malnutrition	32.2%
Steatorrhea & malabsorption	27.2%
Meconium ileus	19.9%
Family history	15.3%
Neonatal screening	4.5%

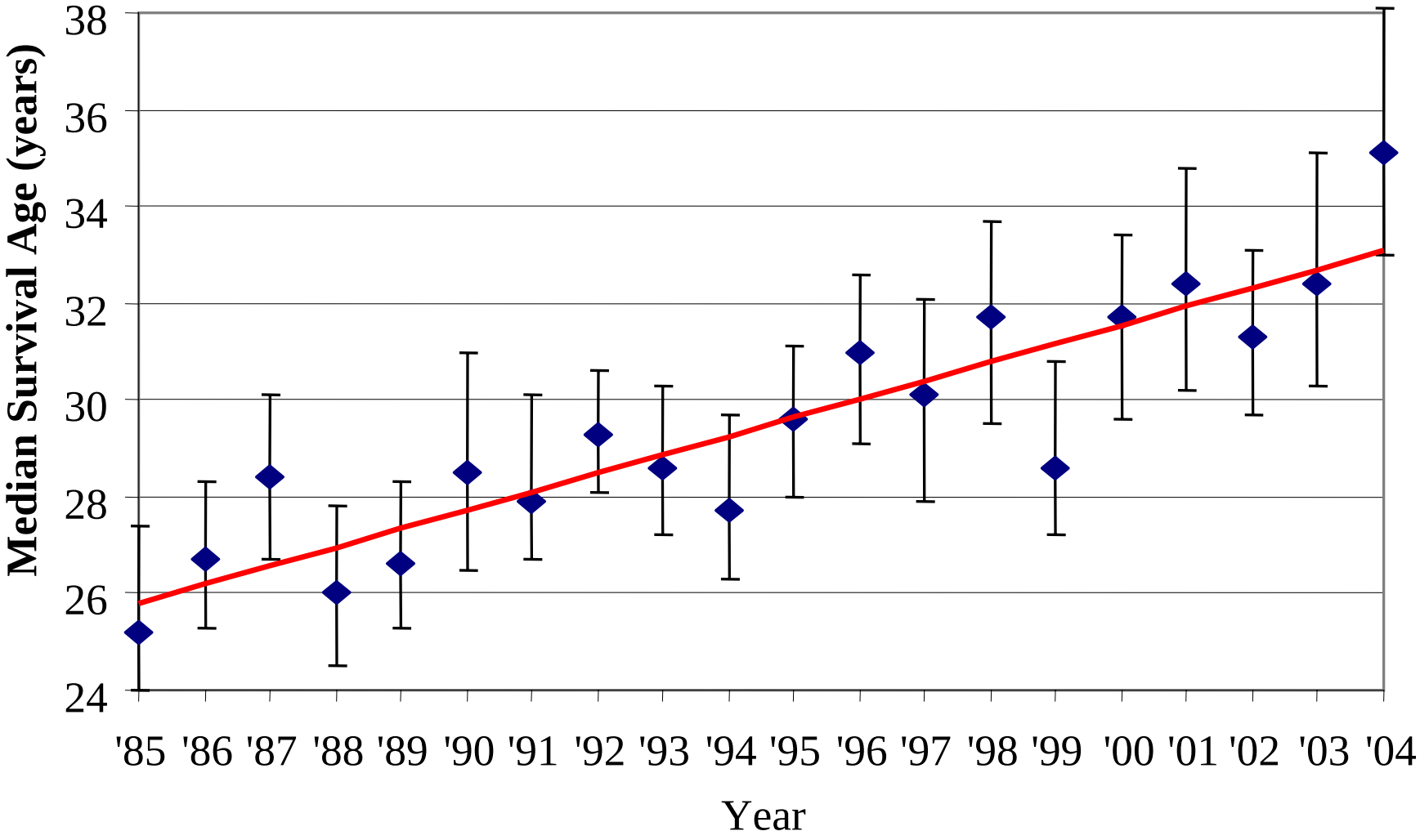
Family History and CF Dx

- Literature estimates - must distinguish between:
  - CF diagnosis because of + FHx
    - Only information is that a relative is affected
    - underwent carrier testing (population scr, infertility, +FHx)
    - underwent prenatal diagnosis
      - screening ultrasound - echogenic or dilated bowel
      - amniocentesis for AMA or specific risks
    - had newborn sweat and or genetic testing done
  - found to have + FHx at time of CF diagnosis
- Together, +FHx estimates were 10 - 15%, but now higher with population screening and +NBS (TP and FP)

Age at Diagnosis - All Patients, 2004

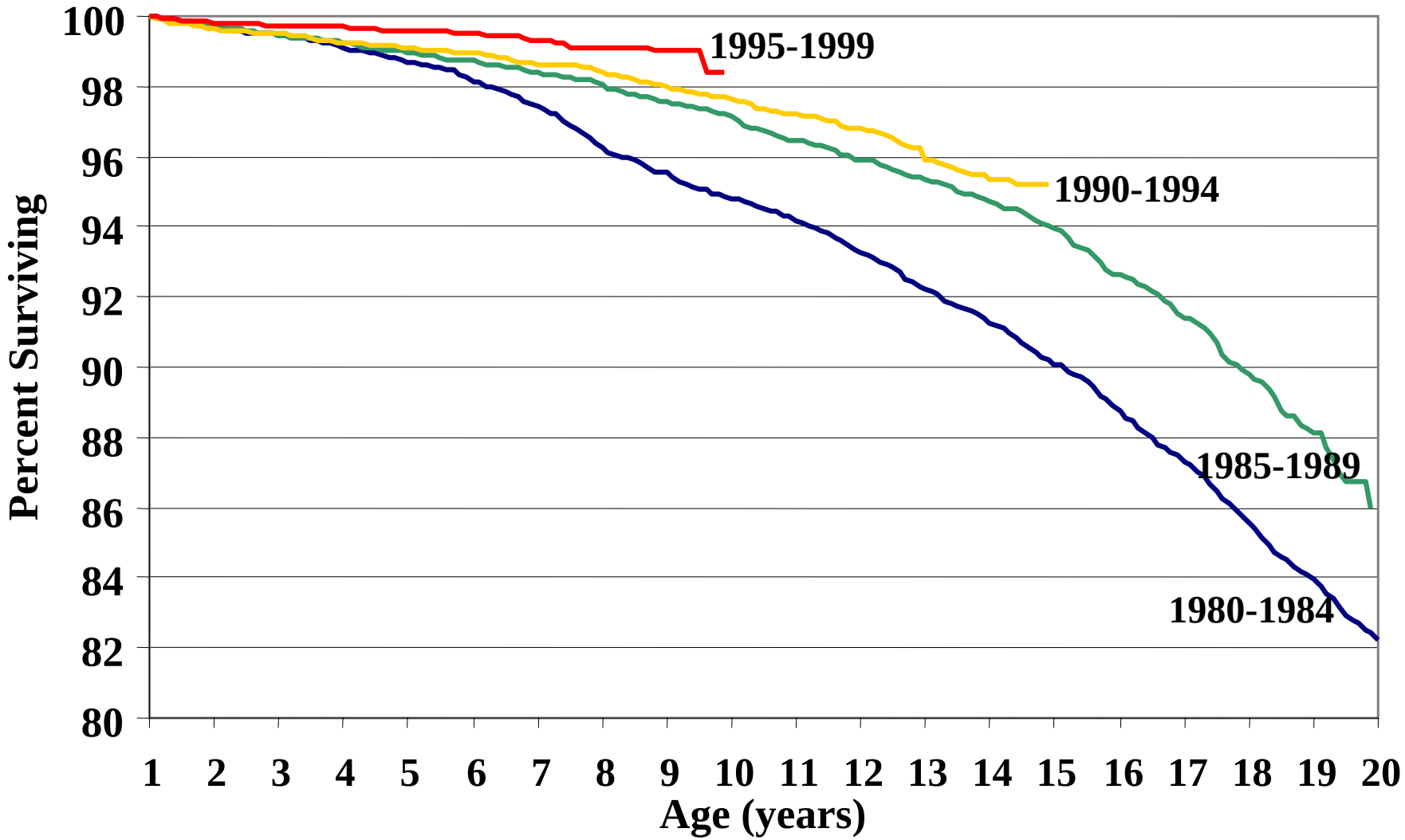


Median Predicted Survival Age, 1985-2004



The median predicted survival is 35.1 years for 2004. The whiskers represent the 95 percent confidence bounds for the survival estimates, so the 2004 median predicted survival is between 33 and 38.1 years.

Survival from Age One, by Year of Birth

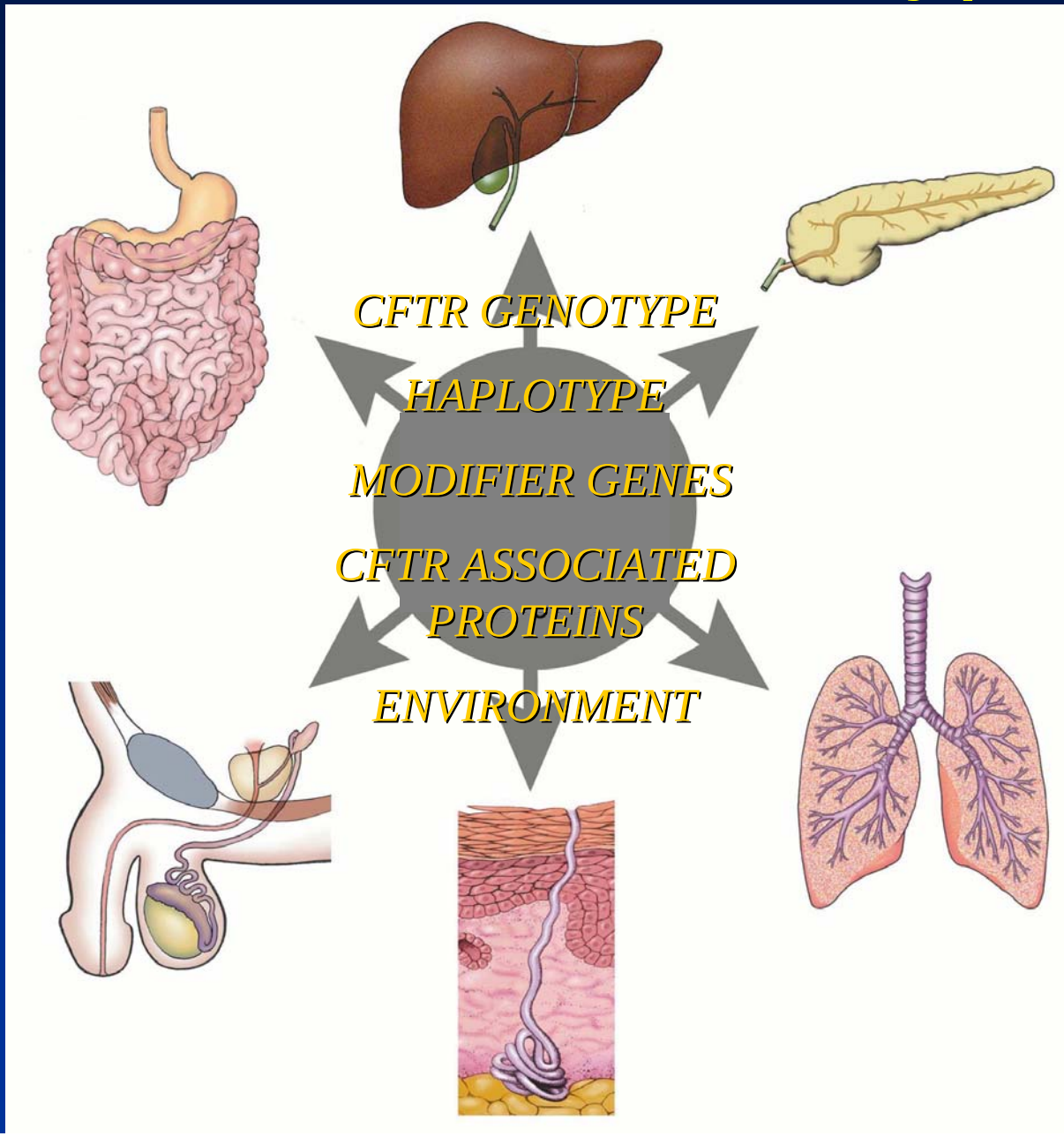


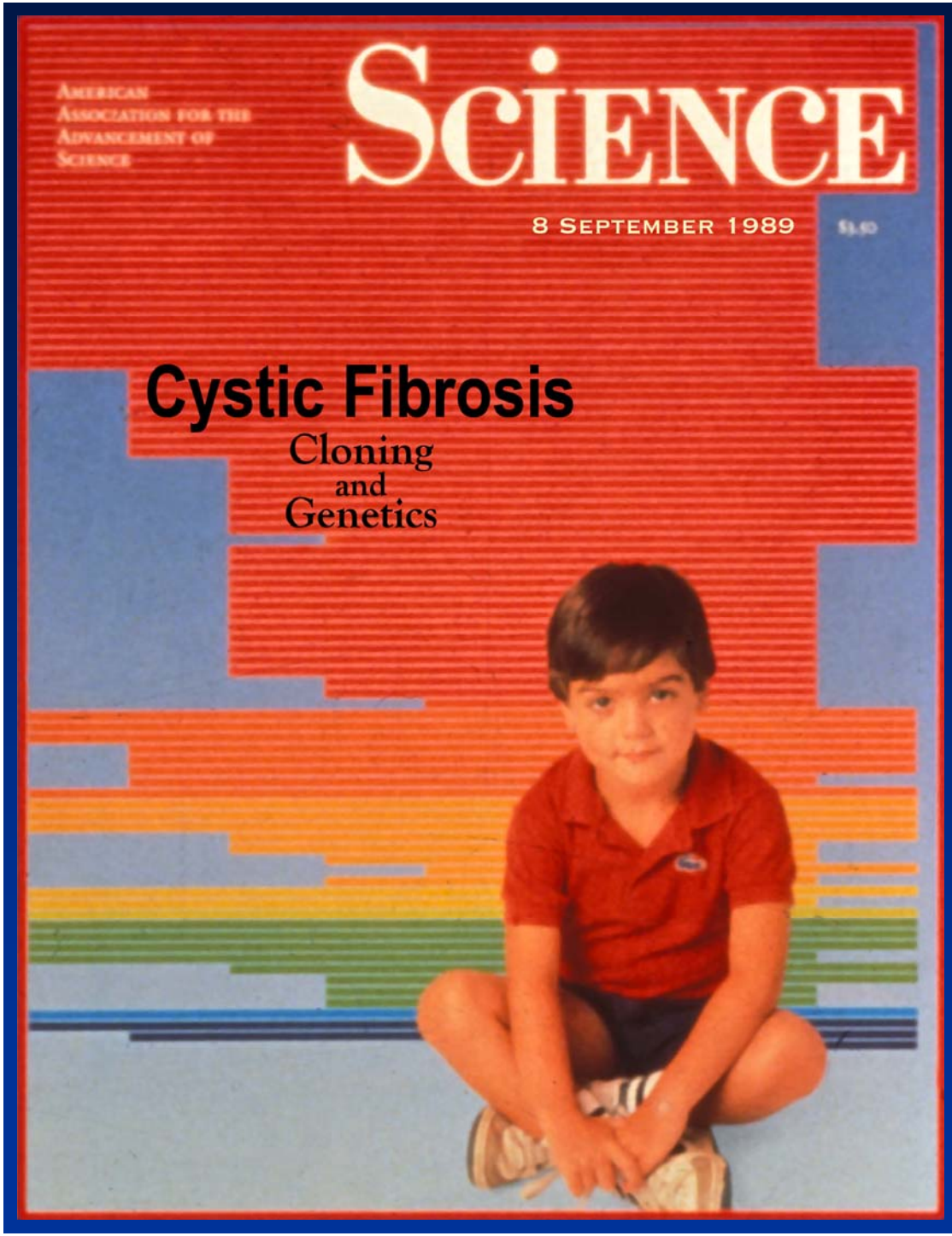
Actual survival of patients in the Registry has steadily improved since 1980. Of patients born between 1980 and 1984 (the earliest cohort shown here), 90.2 percent survived to age 15. For patients born between 1990 and 1994, 95.2 survived to age 15.

Diagnosis

- Phenotype: Lung, GI, GU sx
  - Supporting evidence:
    - Sputum cx, CXR, stool fat, GU exam
- FHx
- CFTR Function:
  - Pilocarpine iontophoresis: sweat [Cl-] 75mg sweat
    - Normal <40 (<30 for newborns)
    - Borderline 40 - 59 (30 - 59 for newborns)
    - CF $\geq$ 60
  - Nasal PD
- Genotype: At least one mutation detected

Modifiers of Phenotype

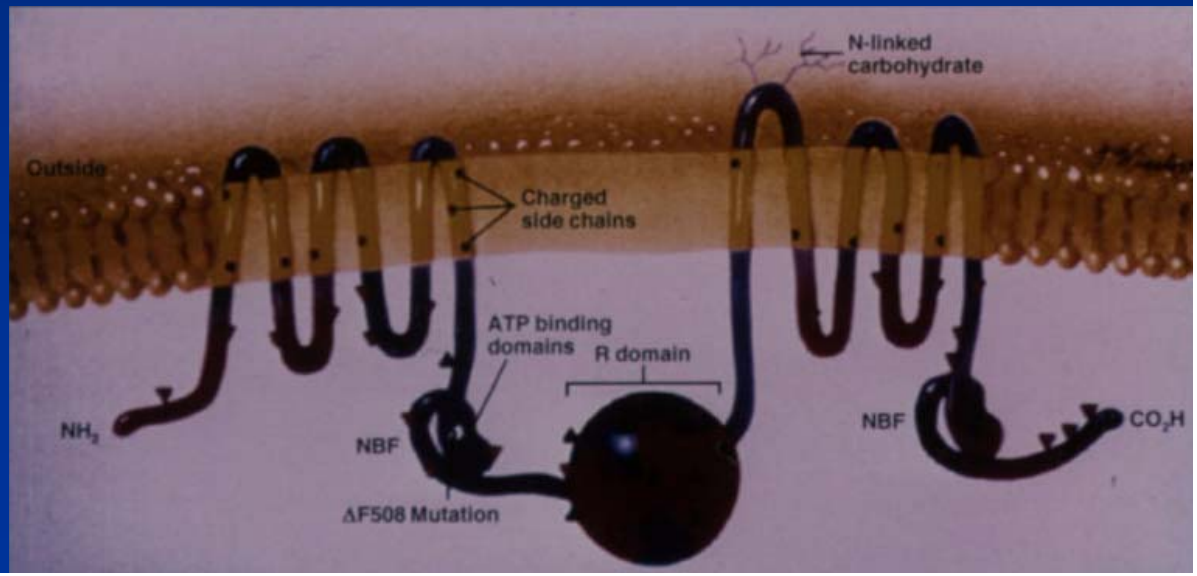




Discovery of CFTR and the ΔF508 Mutation

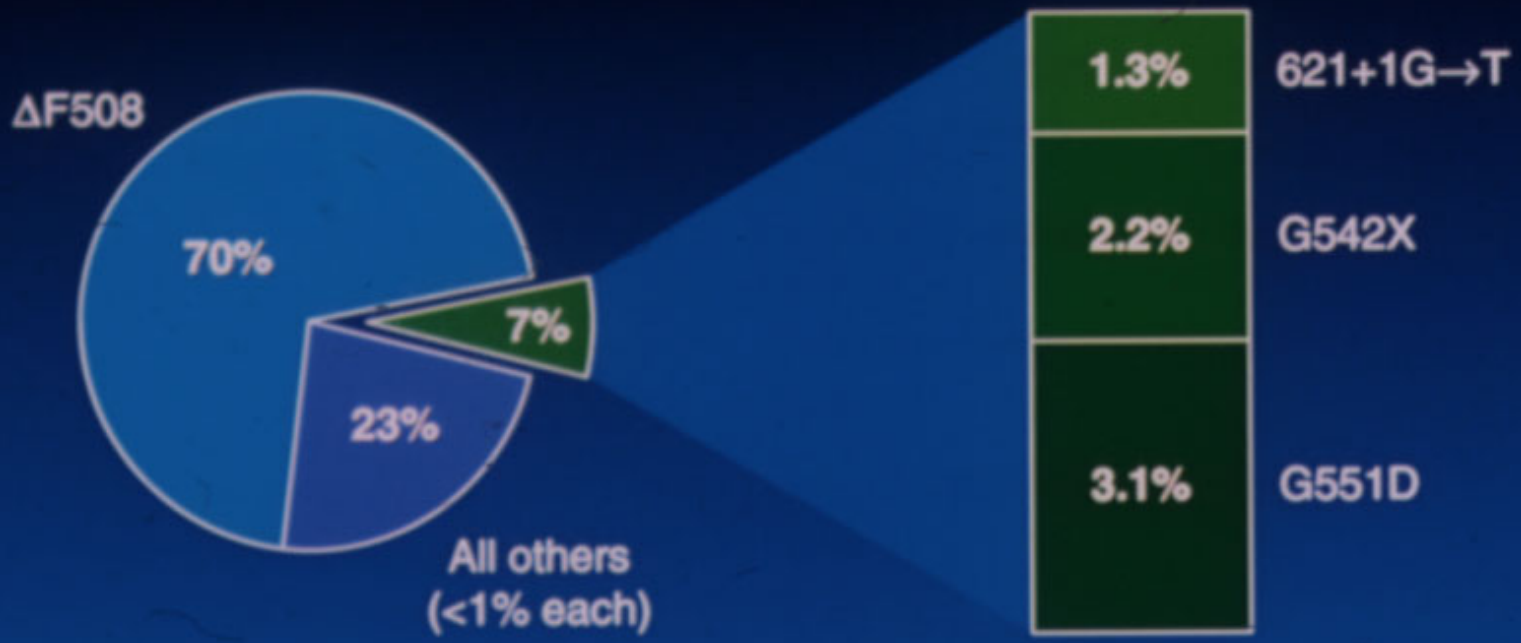
Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) Gene

- Gene 250,000 bp
- mRNA 6,000 bp
- Protein 1480 aa
  - ABC transporter superfamily
- Mutations identified >1,400 (2005)



Frequency of 25 CF Gene Mutations

20 of 25 mutations tested were found (84% of CF alleles)

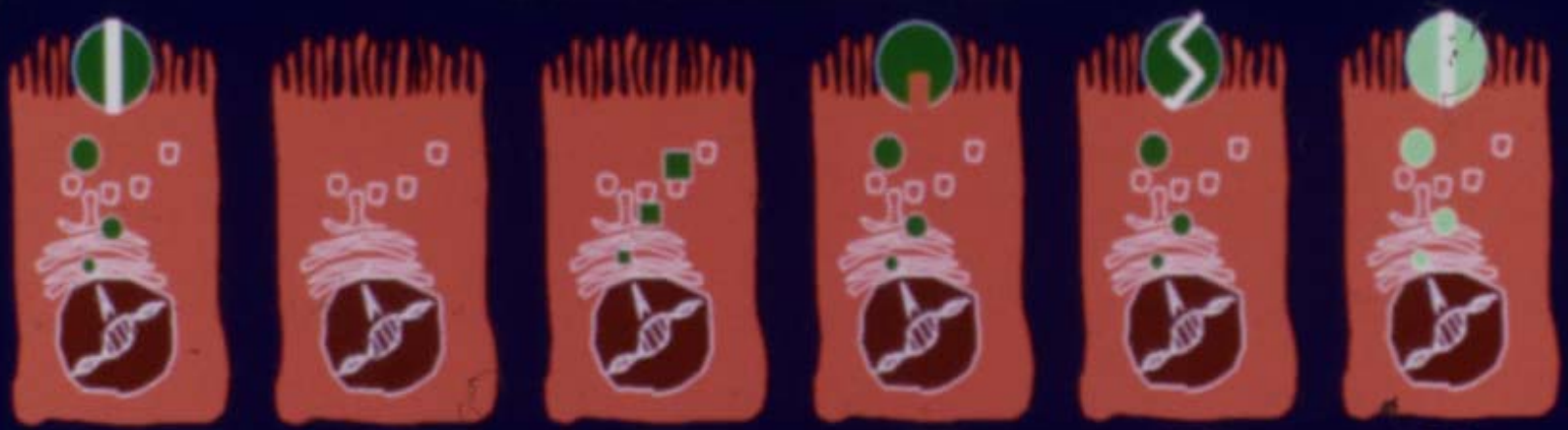


Kristidis P, et al. *Am J Hum Genet.* 1992;50:1178-84.

GEOGRAPHICAL DISTRIBUTION OF F508

	% CF CHROM F508
UK	0.80
Toronto	0.71
Spain	0.51
Italy N/C/S	0.75/0.50/0.30
Israel	0.30
Baltimore(Black)	0.37
Boston	0.62

Molecular Consequences of CFTR Mutations



Normal	I	II	III	IV	V
	No synthesis	Block in processing	Block in regulation	Altered conductance	Reduced synthesis
	Nonsense G542X	Missense	Missense G551D	Missense R117H	Missense A455E
	Frameshift 394delTT	AA deletion ΔF508			Alternative Splicing 3849+10kbC–>T
	Splice junction 1717-1G–>A				

Newborn Screening Impact

Diagnosis as newborn by NBS

3 month old diagnosed
during 2001 in a non-screening state

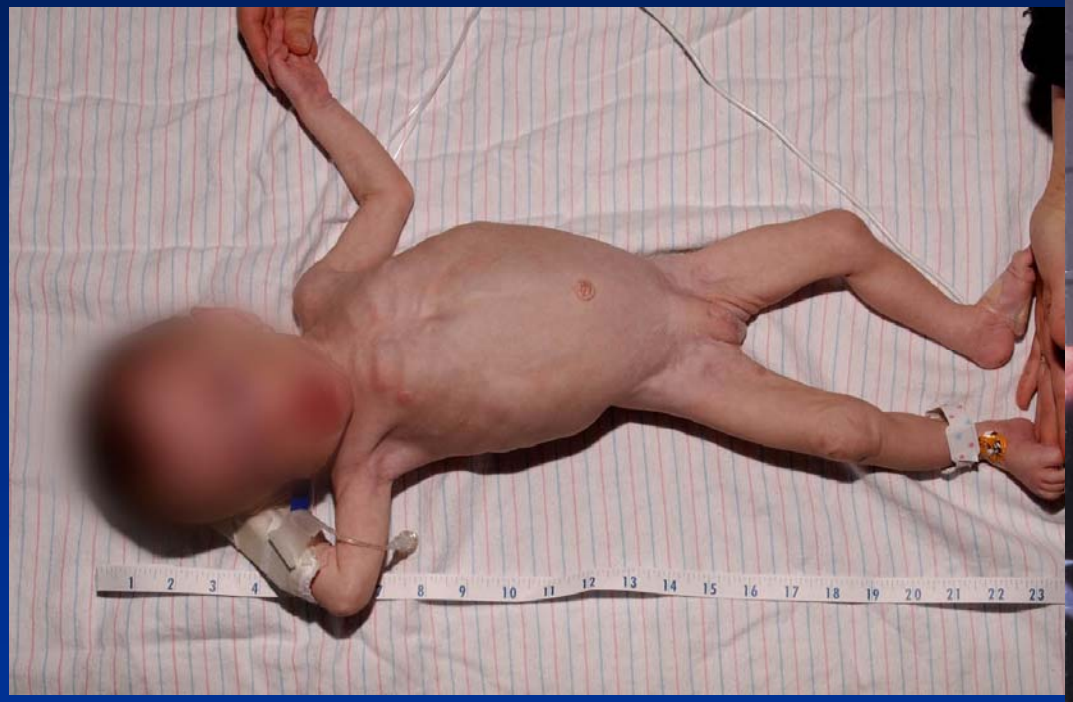
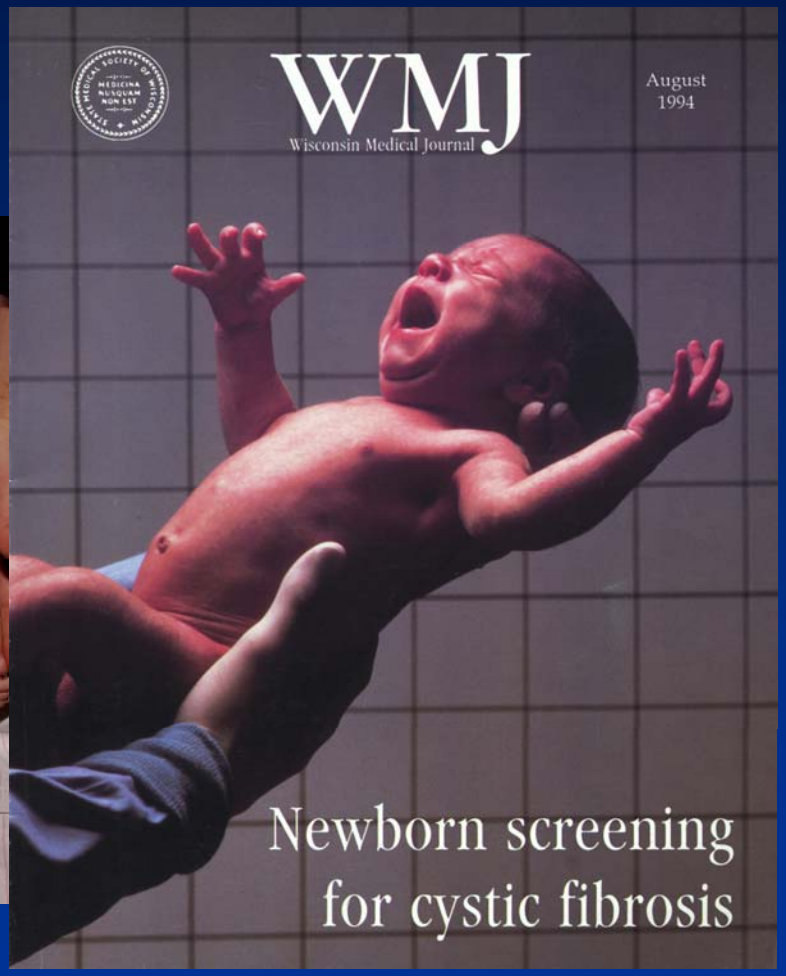
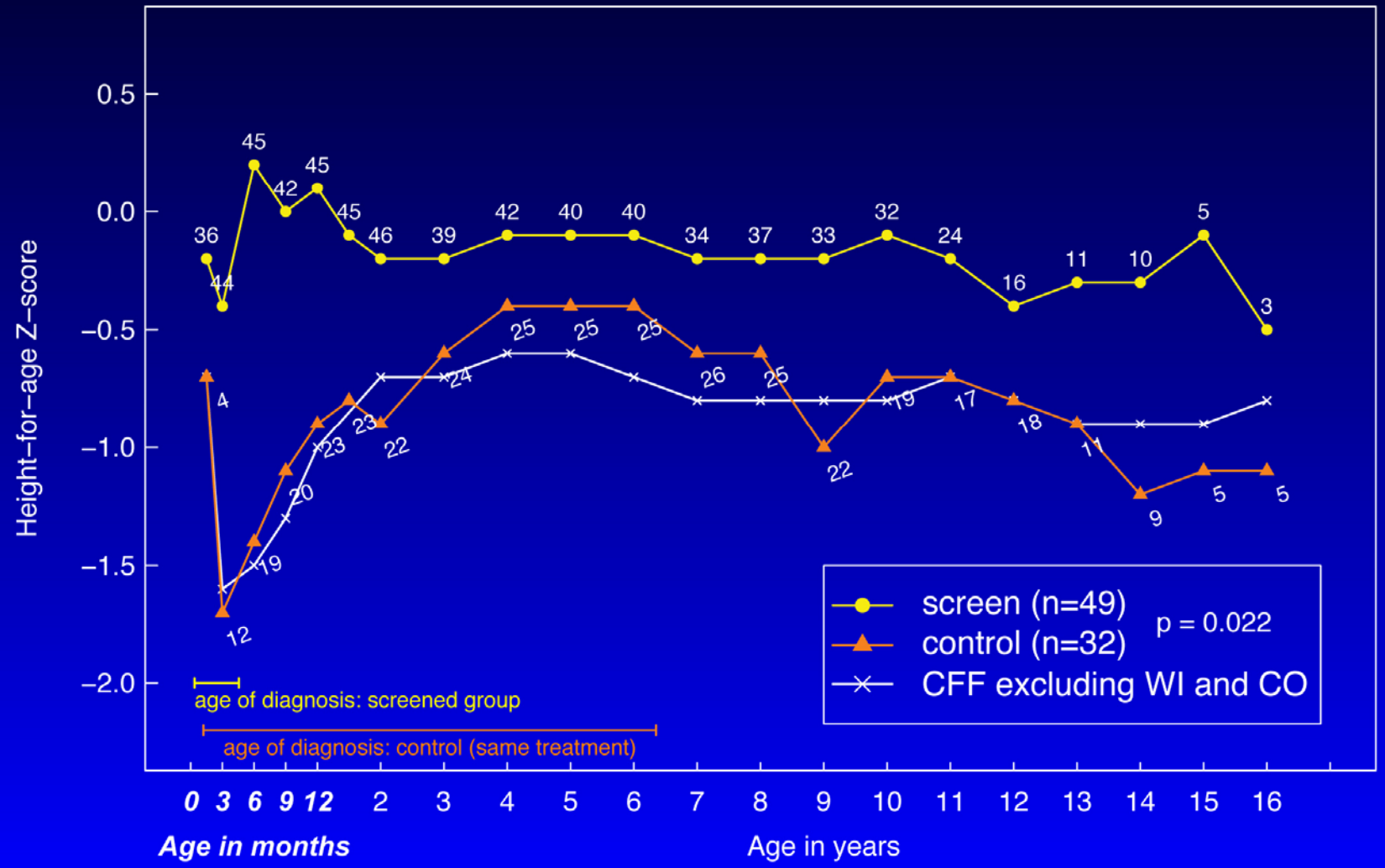


Photo courtesy of Frank J. Accurso, MD



Wisconsin CF Neonatal Screening RCT: Growth of Screened and Traditionally Diagnosed Patients

(Farrell et al, J Pediatrics 2005;147:S30-S36)



CDC

MMWR™

Morbidity and Mortality Weekly Report

Recommendations and Reports | October 15, 2004 / Vol. 53 / No. RR-13

Newborn Screening for Cystic Fibrosis

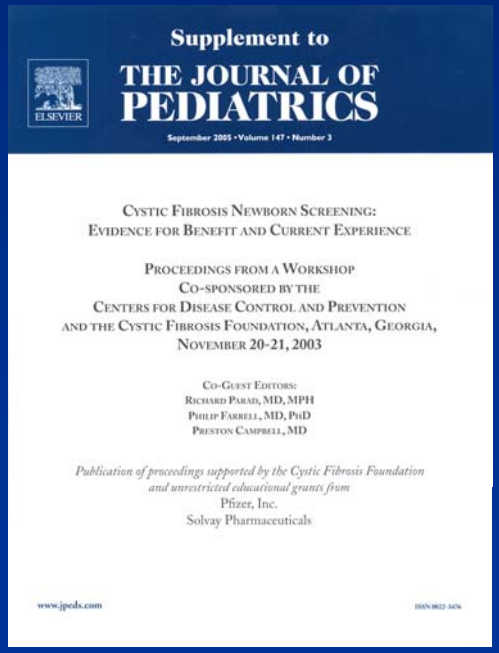
Evaluation of Benefits and Risks and Recommendations for State Newborn Screening Programs

Image courtesy of Natus Medical Incorporated

INSIDE: Continuing Education Examination

DEPARTMENT OF HEALTH AND HUMAN SERVICES
CENTERS FOR DISEASE CONTROL AND PREVENTION

"On the basis of a preponderance of evidence, the health benefits to children with CF outweigh the risk of harm and justify screening for CF."



Supplement to

THE JOURNAL OF PEDIATRICS

September 2005 • Volume 147 • Number 3

CYSTIC FIBROSIS NEWBORN SCREENING:
EVIDENCE FOR BENEFIT AND CURRENT EXPERIENCE

PROCEEDINGS FROM A WORKSHOP
CO-SPONSORED BY THE
CENTERS FOR DISEASE CONTROL AND PREVENTION
AND THE CYSTIC FIBROSIS FOUNDATION, ATLANTA, GEORGIA,
NOVEMBER 20-21, 2003

CO-GUEST EDITORS:
RICHARD PARAD, MD, MPH
PHILIP FARRELL, MD, PhD
PRESTON CAMPBELL, MD

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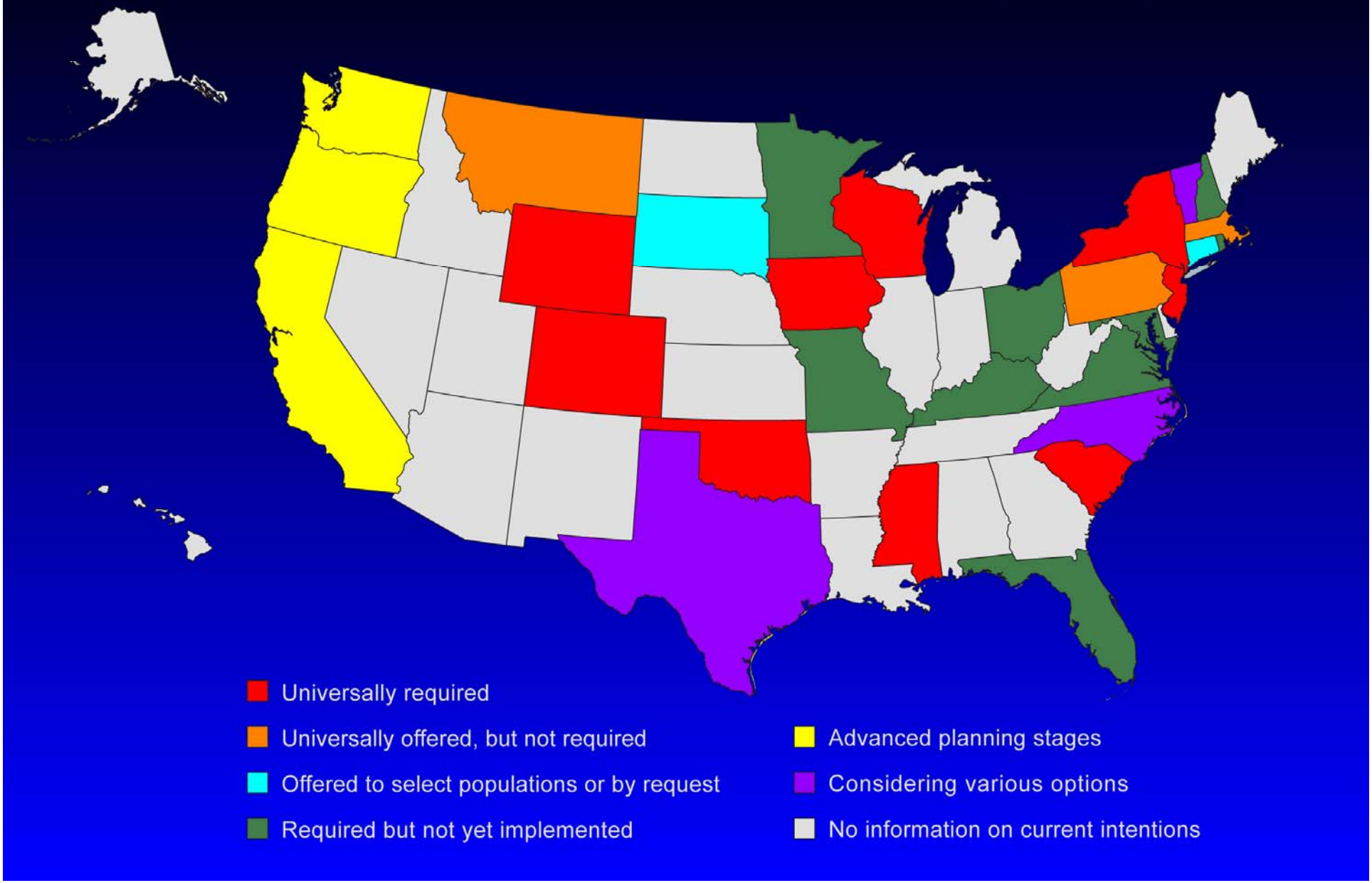
www.jpeds.com

Current Status of CF NBS (2006)



Projected Status of CF Newborn Screening

US annual births ~ 4,000,000

	% births screened	New diagnoses
2000	5%	50
2006	25%	250
2007	70%	700
2010 (CFF target)	100%	1,000

Population Screening: NIH Consensus Statement 1997

- Offer CF genetic screening to:
  - Adults with positive FHx CF
  - Partners of CF affected
  - Couples planning pregnancy
  - Couples seeking prenatal care
- ?Implementation

Population Screening

PRO

- Maximize parental options
- Prevention of affecteds
- ?Cost effective

CON

- Test is imperfect
- Test is difficult to understand
- Results cause anxiety
- Parents may not act on results (25-75% terminate)

ACMG/ACOG/NIH

- Implementation issues discussed:
  - target population (universal vs. high risk ethnicities)
  - couple vs. sequential
  - mutation panel selection
  - extended testing
  - mild variant mutations
  - test interpretation, reporting , genetic counseling
  - laboratory QA

Joint recommendations for preconceptual and prenatal carrier screening for CF

- Parent education pamphlets
- Provider education programs
- Who prepares/consents parents?
- Who reports genotype?
- Who is referred to genetic counseling?

ACMG 25

ΔF508	G551D	R117H	W1282X	2789+5G>A
ΔI507	G85E	R334W	1078delT	3120G>A
A455E	I148T	R347P	1717-1G>A	3659delC
711+1G>T	N1303K	R553X	1898+1G>A	3849+10kbC>T
G542X	R1162X	621+1 G>T	2184delA	621+1G>T

Risk after carrier testing

% mutations detected	Carrier risk if -	Risk CF offspring one parent +	Risk CF both parents -
0	1/25	NA	1/2500
70	1/83	1/331	1/27,000
80	1/124	1/494	1/61,000
85	1/165	1/661	1/109,200
90	1/246	1/984	1,242,100
95	1/491	1/1964	1/964,200

Ethnic adjustment for carrier risk

		Estimated carrier risk	
Ethnic group	**Detection rate**	**Before test**	**After negative test**
Ashkenazi Jewish	97%	1/29	~1/930
European Caucasian	80%	1/29	~1/140
African American	69%	1/65	~1/207
Hispanic American	57%	1/46	~1/105
Asian American	-	1/90	-

What do patients need to understand before consenting to testing?

- What is the disease and what is its outcome - e.g., median survival, is there a cure?
- What are the Mendelian genetics ?
- What are the weaknesses of the genetic test (false negative)
- What will happen if the test is positive (what are the options for termination, insurance implications, clinical status of carrier)

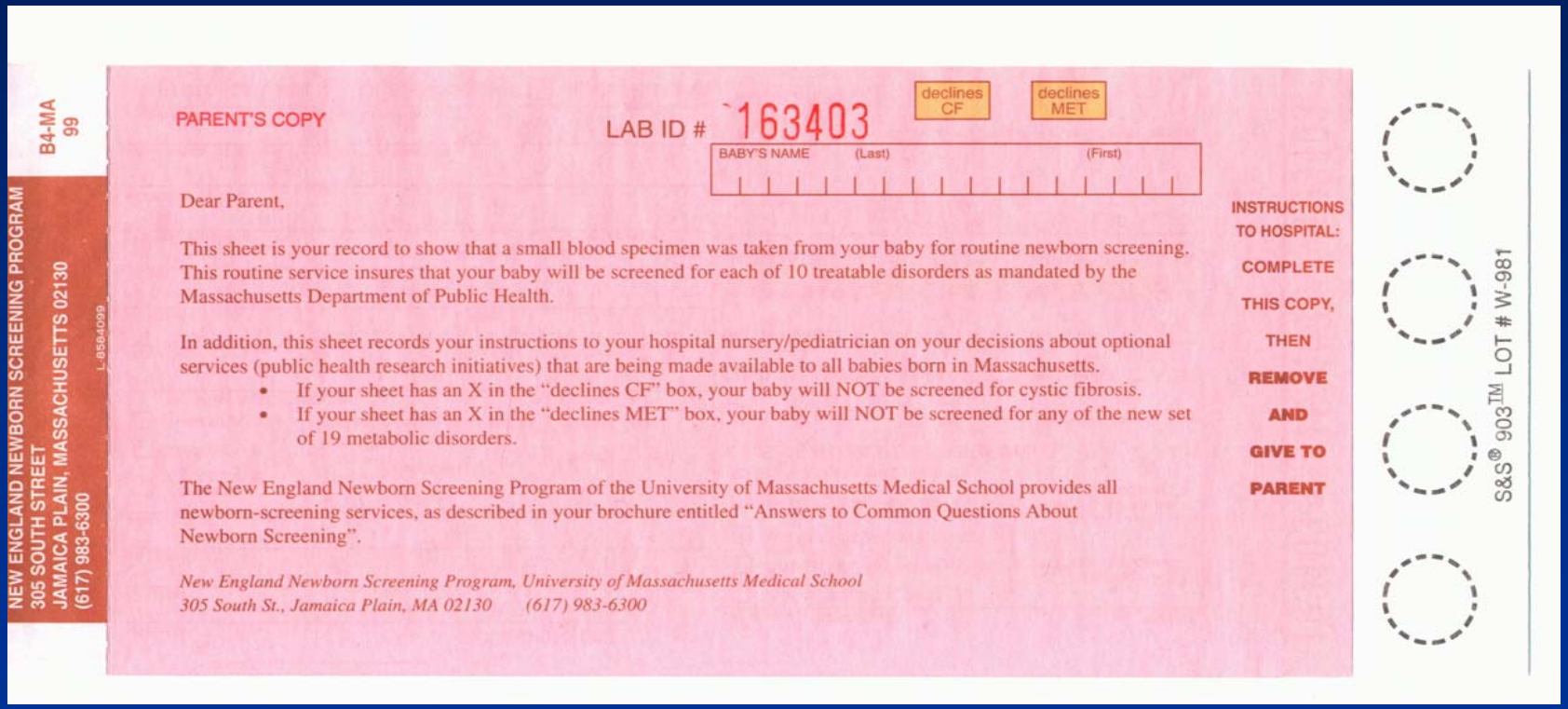
Cystic Fibrosis Carrier Testing:

The Decision Is Yours

Cystic Fibrosis Testing:

What Happens If Both My Partner and I Are Carriers?

Massachusetts Newborn Screening for CF



B4-MA
99

NEW ENGLAND NEWBORN SCREENING PROGRAM
305 SOUTH STREET
JAMAICA PLAIN, MASSACHUSETTS 02130
(617) 983-6300

L-858-4099

PARENT'S COPY

LAB ID # 163403

declines CF

declines MET

BABY'S NAME (Last) (First)

Dear Parent,

This sheet is your record to show that a small blood specimen was taken from your baby for routine newborn screening. This routine service insures that your baby will be screened for each of 10 treatable disorders as mandated by the Massachusetts Department of Public Health.

In addition, this sheet records your instructions to your hospital nursery/pediatrician on your decisions about optional services (public health research initiatives) that are being made available to all babies born in Massachusetts.

- If your sheet has an X in the "declines CF" box, your baby will NOT be screened for cystic fibrosis.
- If your sheet has an X in the "declines MET" box, your baby will NOT be screened for any of the new set of 19 metabolic disorders.

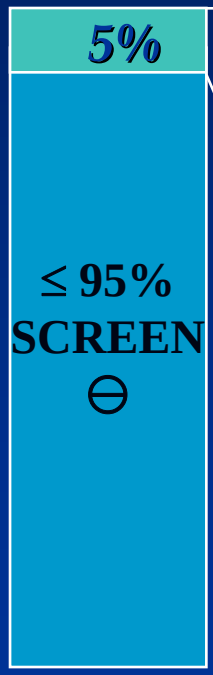
The New England Newborn Screening Program of the University of Massachusetts Medical School provides all newborn-screening services, as described in your brochure entitled "Answers to Common Questions About Newborn Screening".

New England Newborn Screening Program, University of Massachusetts Medical School
305 South St., Jamaica Plain, MA 02130 (617) 983-6300

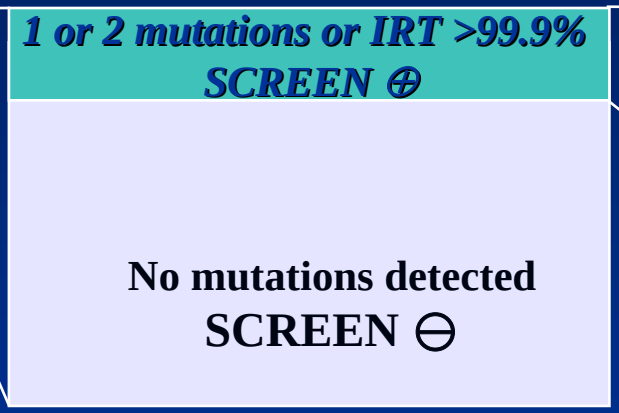
INSTRUCTIONS TO HOSPITAL: COMPLETE THIS COPY, THEN REMOVE AND GIVE TO PARENT

S&S® 903™ LOT # W-981

Massachusetts CF Newborn Screen IRT/DNA Algorithm



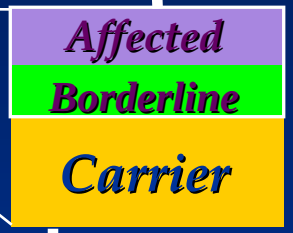
IRT



DNA

39* MUTATION PANEL

*(was 16,27, 25)



CF Specialist

SWEAT CHLORIDE

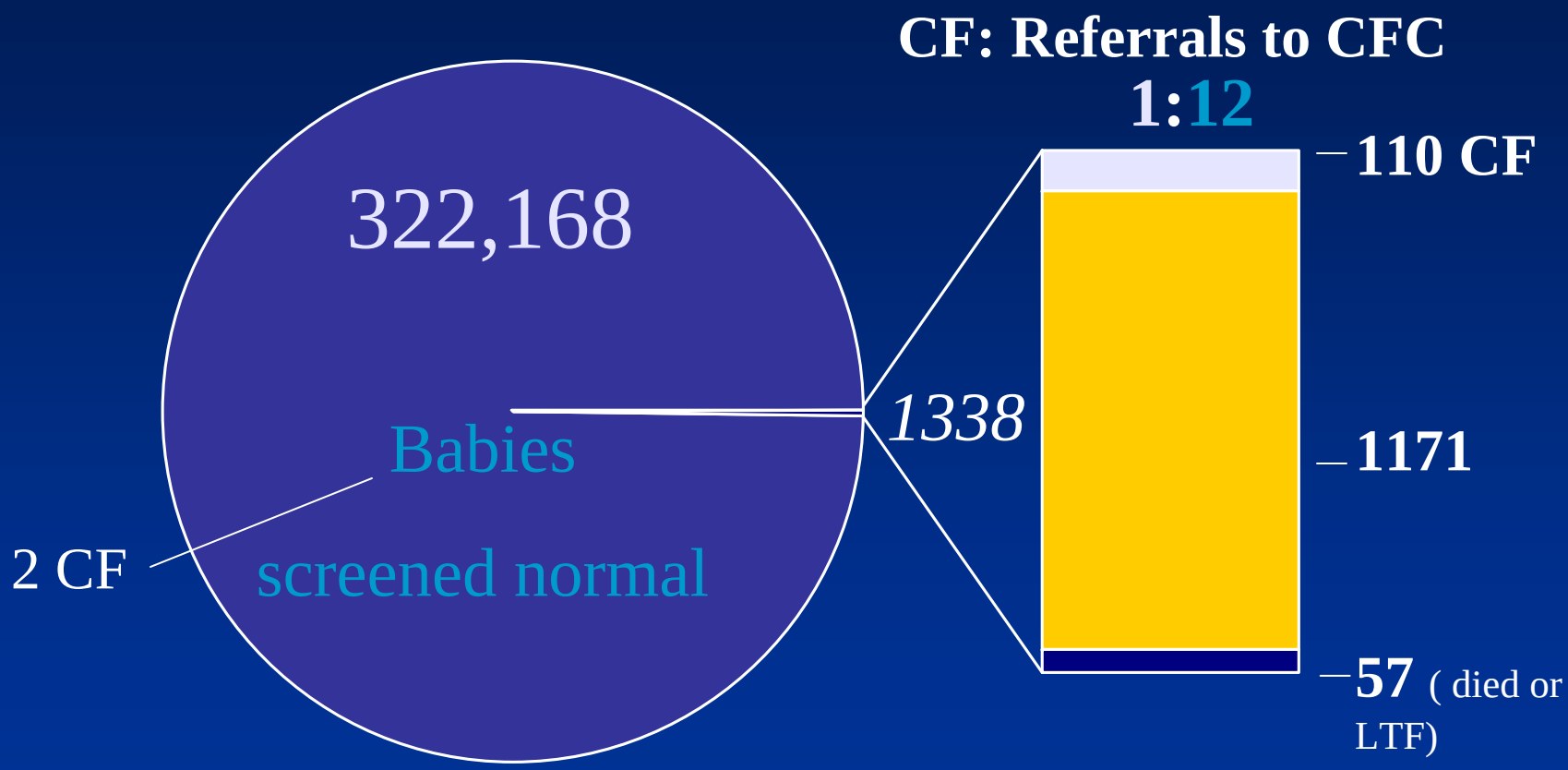
Genetic Counselor

*ΔF508, R117H, G551D, G542X, W1282X, N1303K, R334W, 621+1G>T, R553X, Δ I507, 1717-1G>A, R347P, R560T, 3849+10kbC>T, A455E, 3120+1G>A, 3659delC, A559T, R1162X, S1255X, 405+3A>C, 711+1G>T, 2789+5G>A, G480C, 2307insA, G85E , 1078delT

Contact Algorithm: Call to Pedi with collection of FHx

- C elevated IRT, 2 CFTR mutations (6%)
  - 1:1 likelihood of CF
  - refer to CF center for intake/sweat test
- B elevated IRT, 1 CFTR mutation (70%)
  - 1:40 likelihood of CF: likely carrier
  - refer for sweat testing, genetic counseling
  - emphasis on genetic counseling post-sweat result
- A elevated IRT, zero CFTR mutations (24%)
  - 1:100 likelihood of CF: probable FP. ↑ICU, ↑ethnic
  - refer for sweat test, no genetic counseling

Yield from Cystic Fibrosis Pilot Screening
Feb 99 through Jan 03



PEDIATRICS Vol. 113 No. 6 June 2004

New England Newborn Screening Program

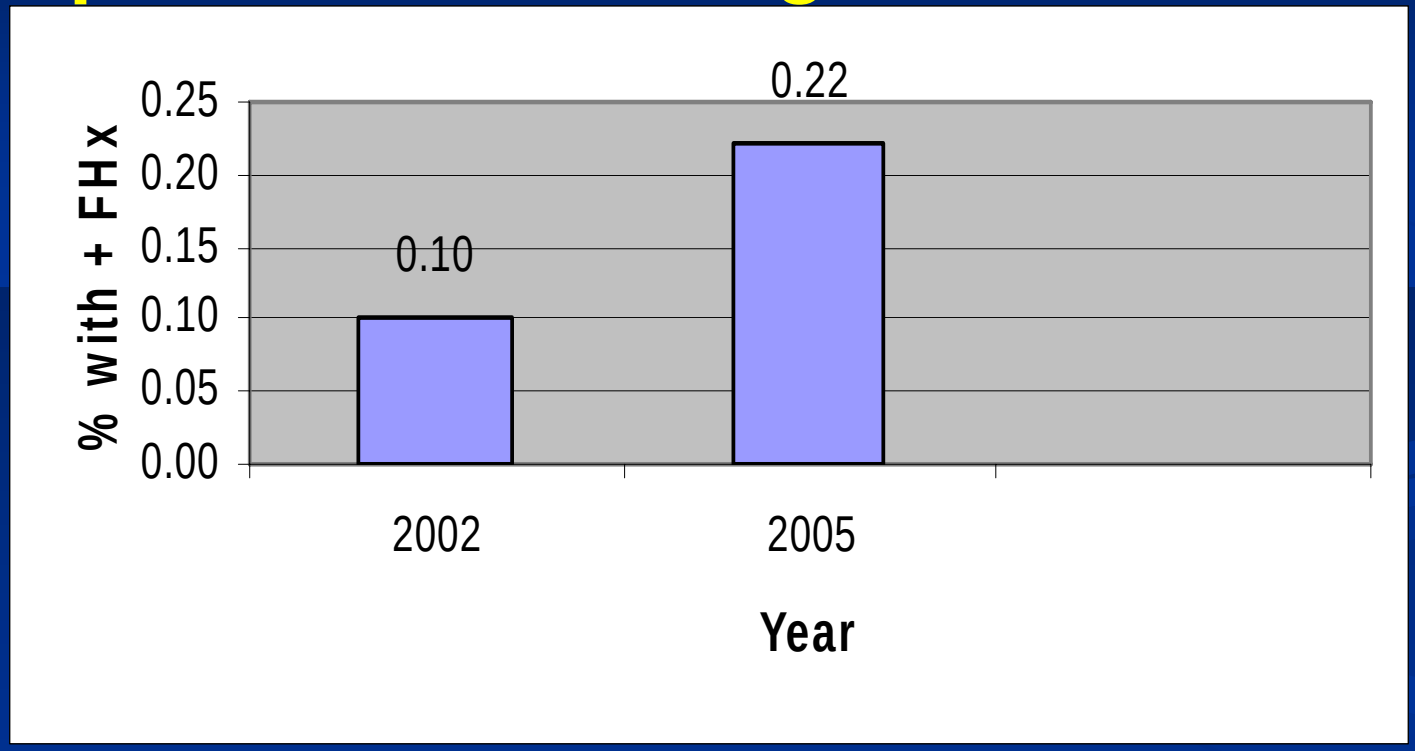
TABLE 2. First Signs Prompting Clinical Suspicion for CF in 110 Screened-Positive, CF-Affected Infants as Reported by Primary Care Provider Upon Receipt of CF-Screen-Positive Result

	n	%
Specific Signs	34	31
Prenatal diagnosis	12	11
Meconium ileus	10	9
Other bowel obstruction	8	7
Both parents carriers; no prenatal diagnosis	4	4
Nonspecific signs (asymptomatic)	22	20
Slow weight gain; stooling issues	17	15
Rule out NEC	1	1
Fever and respiratory symptoms with hospital admission	1	1
Other unrelated*	3	3
Well or no signs prompting clinical suspicion	54	49

NEC indicates necrotizing enterocolitis.
* Transposition of the great arteries, umbilical granuloma, ear malformation.

Increase in % +FHx for CF Carrier

(by Population screening of +NBS in family)



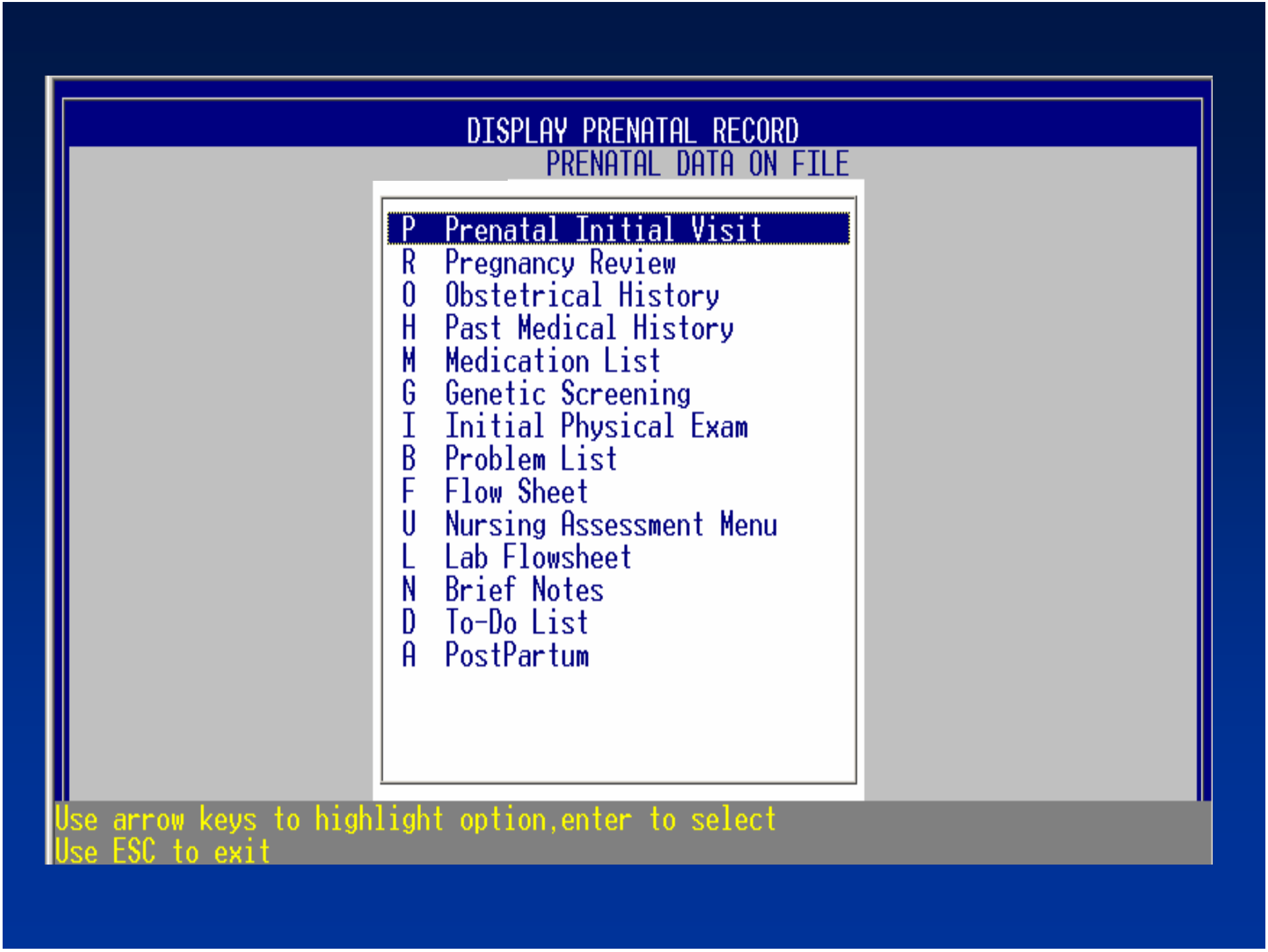
Over 4 years, 325 newborns were referred to a single CF center for +NBS and underwent GC (confirmation of NBS phone hx). 52 (16.7%) had +FHx due to genetic testing in family(population screening or positive newborn screen). Only 2% had affected family members. Note that only 10% of carriers are detected as CFNBS FP.

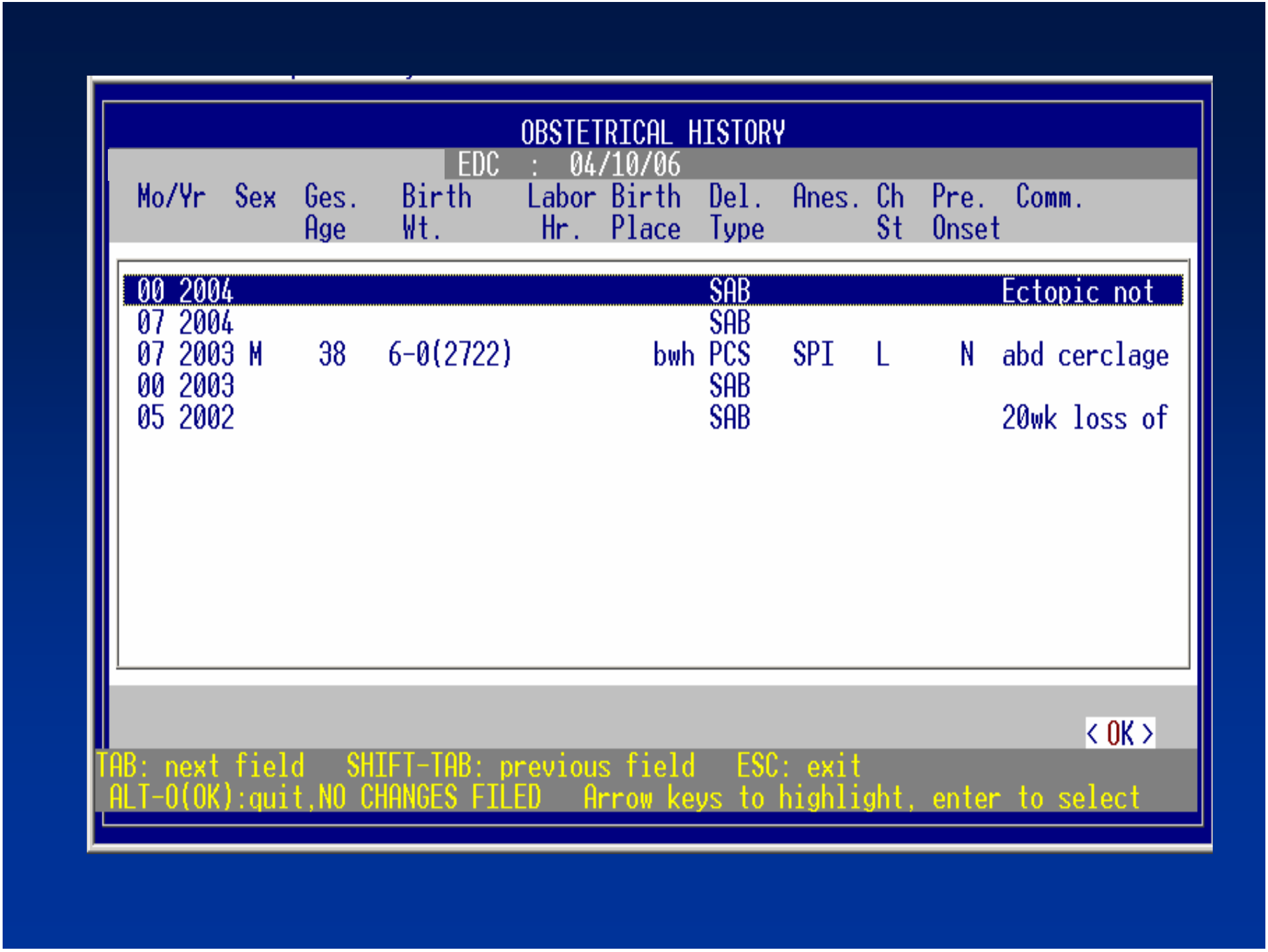
Understanding of Genetics

- Mail survey of 64 CFNBS FP who underwent GC
- Knowledge retention
  - 32% answered all 6 questions correctly
  - 50% answered 5 of 6 questions correctly
- Comprehension among “Non-Carrier” parents
  - Asked to chose between:
    - A. I am definitely not a carrier of CF, or
    - B. There is a small chance that I am a CF carrier
  - 53% answered A, 47% answered B
- 98% of families shared information with other family members
- Anxiety scale showed 2-fold increase in carrier vs. non-carrier

Perspectives

- **Screener**: Many pediatricians, when notified of a +CFNBS result, have not been told by the mother either that she underwent negative CF carrier screening (which ↓ risk), that a mutation was detected or what mutation.
- **Neonatologist**: There is a significant degradation of important medical and family history information at the time of transmission between the mother's record and the newborn's new medical record.


DISPLAY PRENATAL RECORD
PRENATAL DATA ON FILE
P Prenatal Initial Visit
R Pregnancy Review
O Obstetrical History
H Past Medical History
M Medication List
G Genetic Screening
I Initial Physical Exam
B Problem List
F Flow Sheet
U Nursing Assessment Menu
L Lab Flowsheet
N Brief Notes
D To-Do List
A PostPartum
Use arrow keys to highlight option,enter to select
Use ESC to exit


OBSTETRICAL HISTORY
EDC : 04/10/06
Mo/Yr Sex Ges. Age Birth Wt. Labor Hr. Birth Place Del. Type Anes. Ch St Pre. Onset Comm.
00 2004 SAB Ectopic not
07 2004 SAB
07 2003 M 38 6-0(2722) bwh PCS SPI L N abd cerclage
00 2003 SAB
05 2002 SAB 20wk loss of
< OK >
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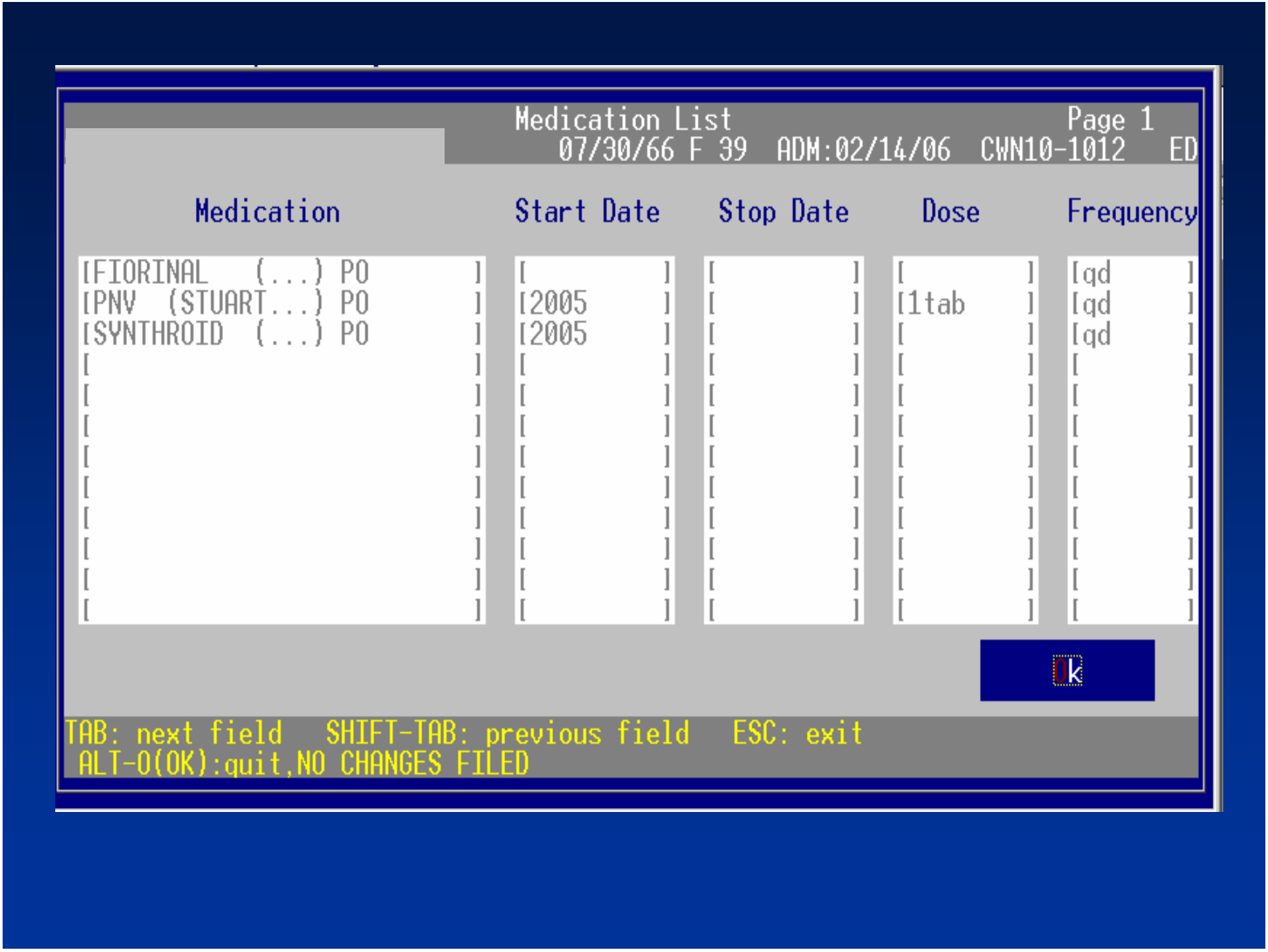
Medication List
Page 1
07/30/66 F 39 ADM:02/14/06 CWN10-1012 ED
Medication
Start Date
Stop Date
Dose
Frequency
[FIORINAL (...) PO]
[PNV (STUART...) PO]
[SYNTHROID (...) PO]
[2005]
[2005]
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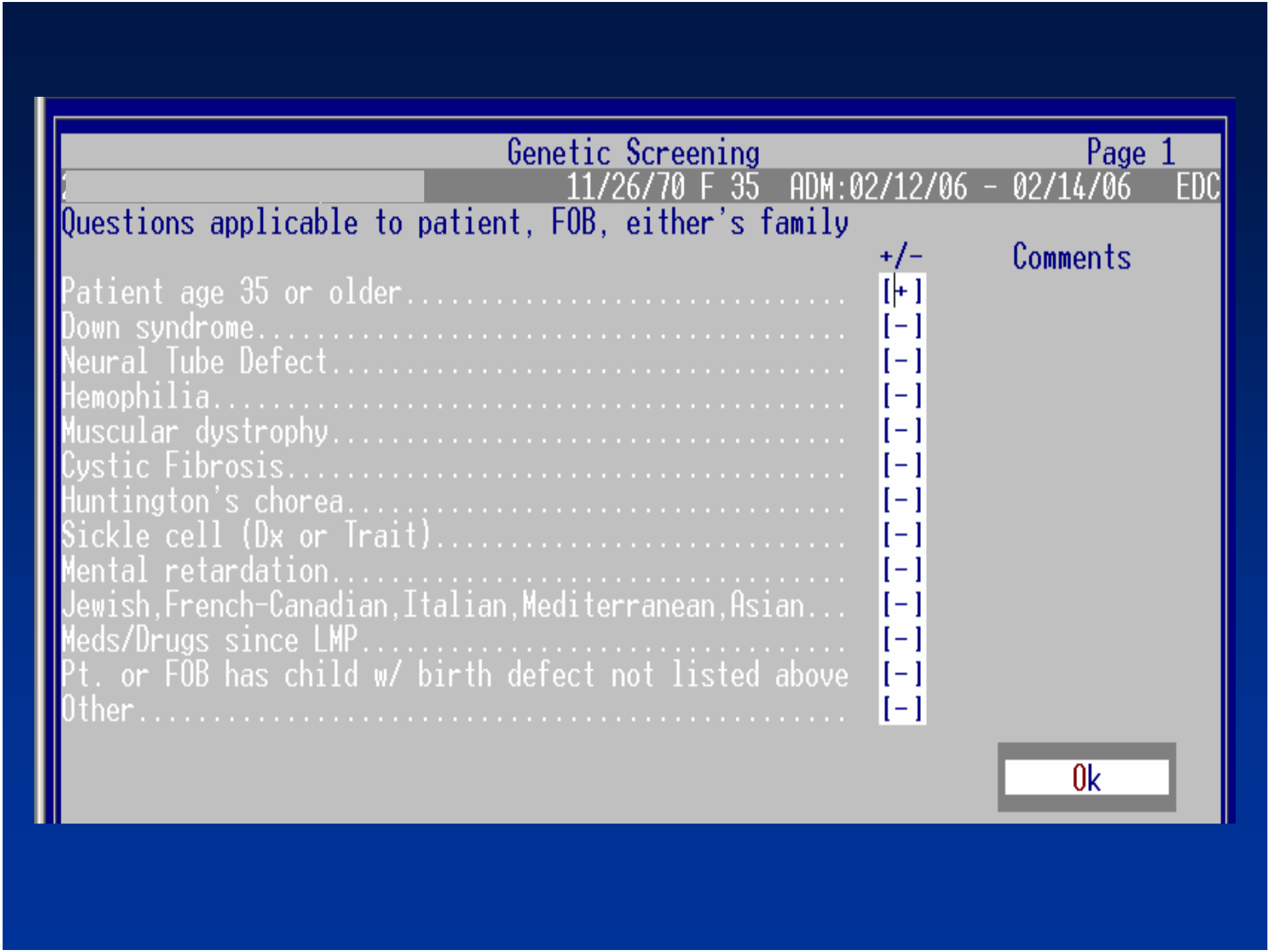
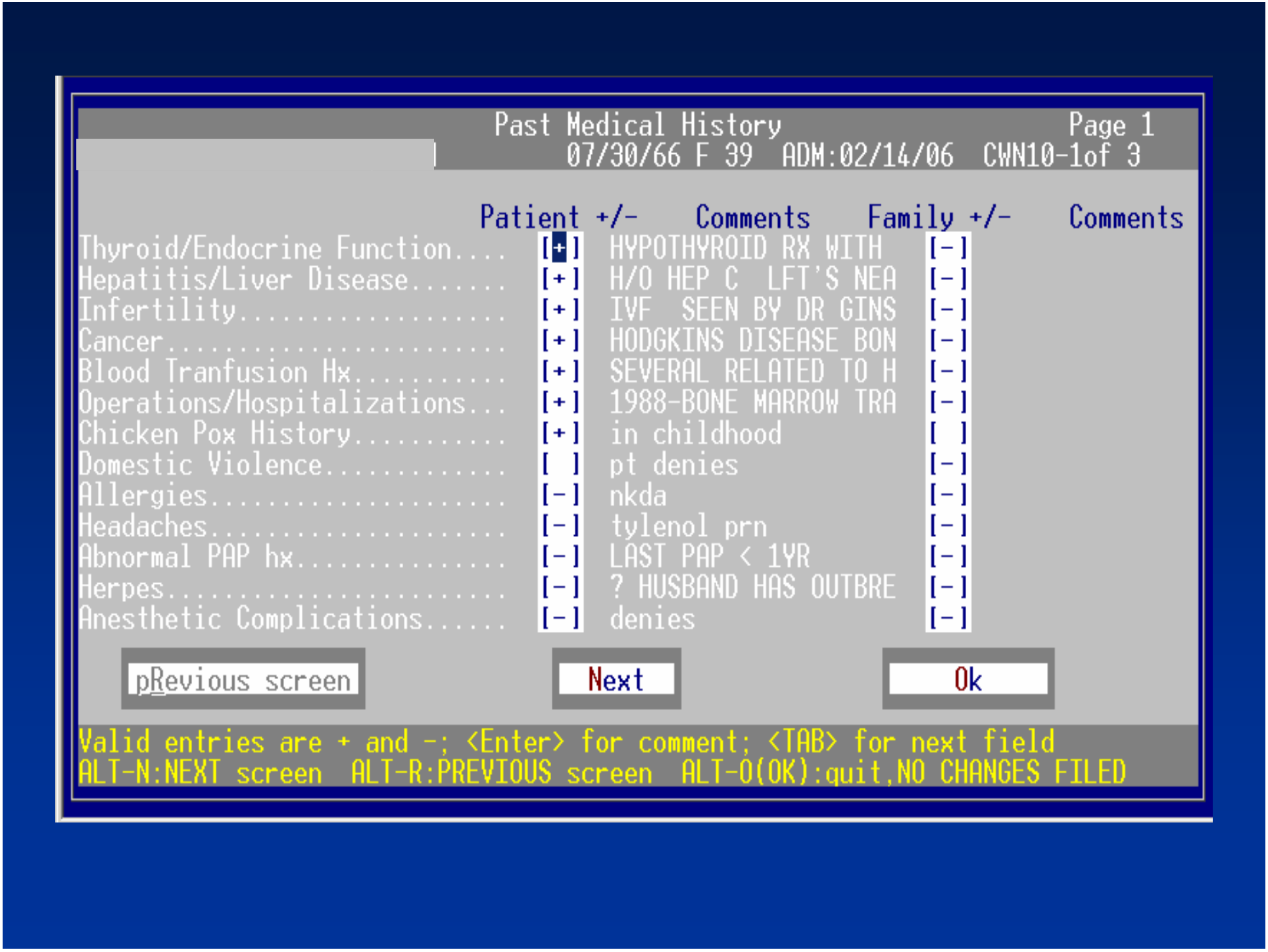
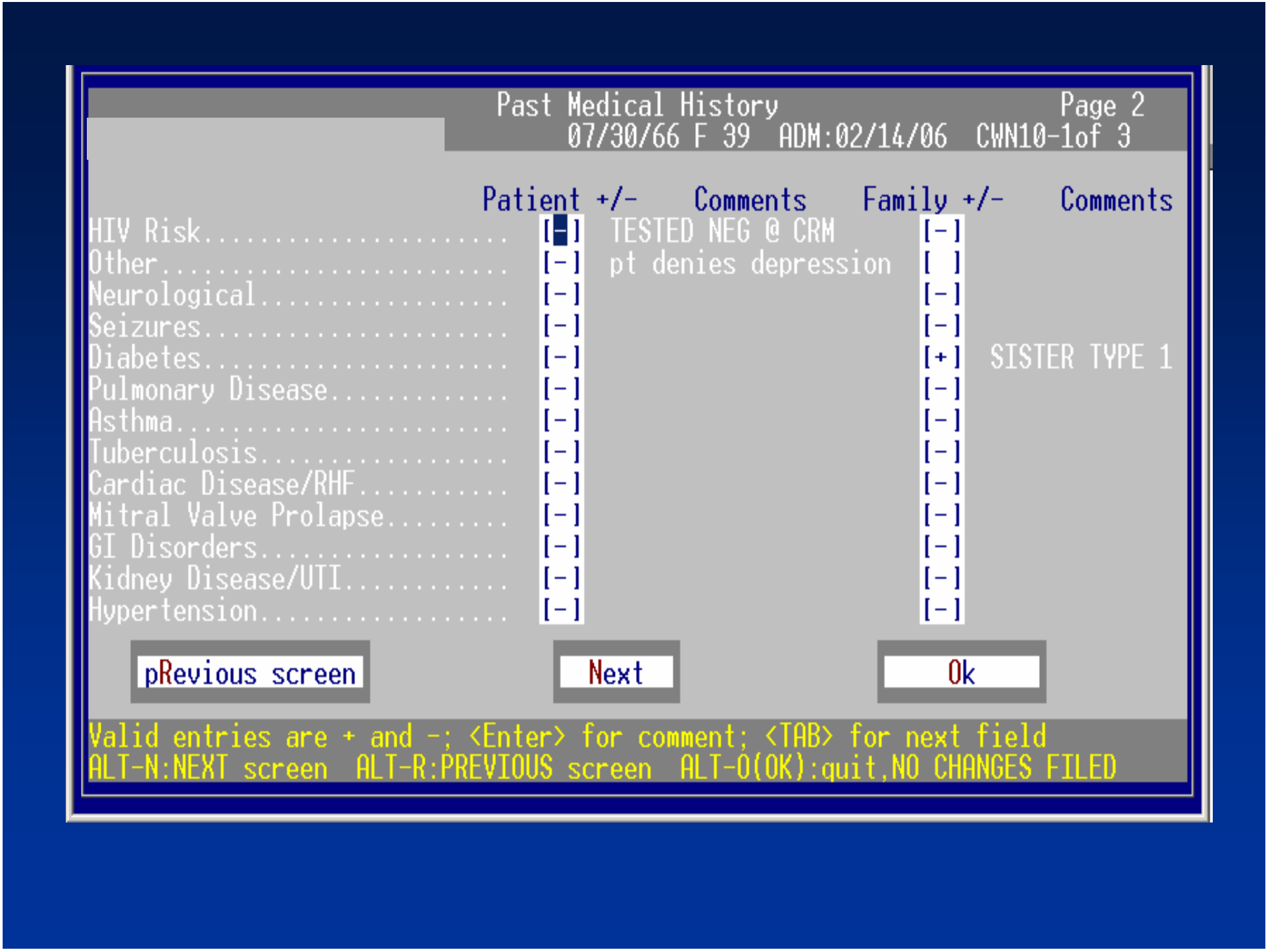
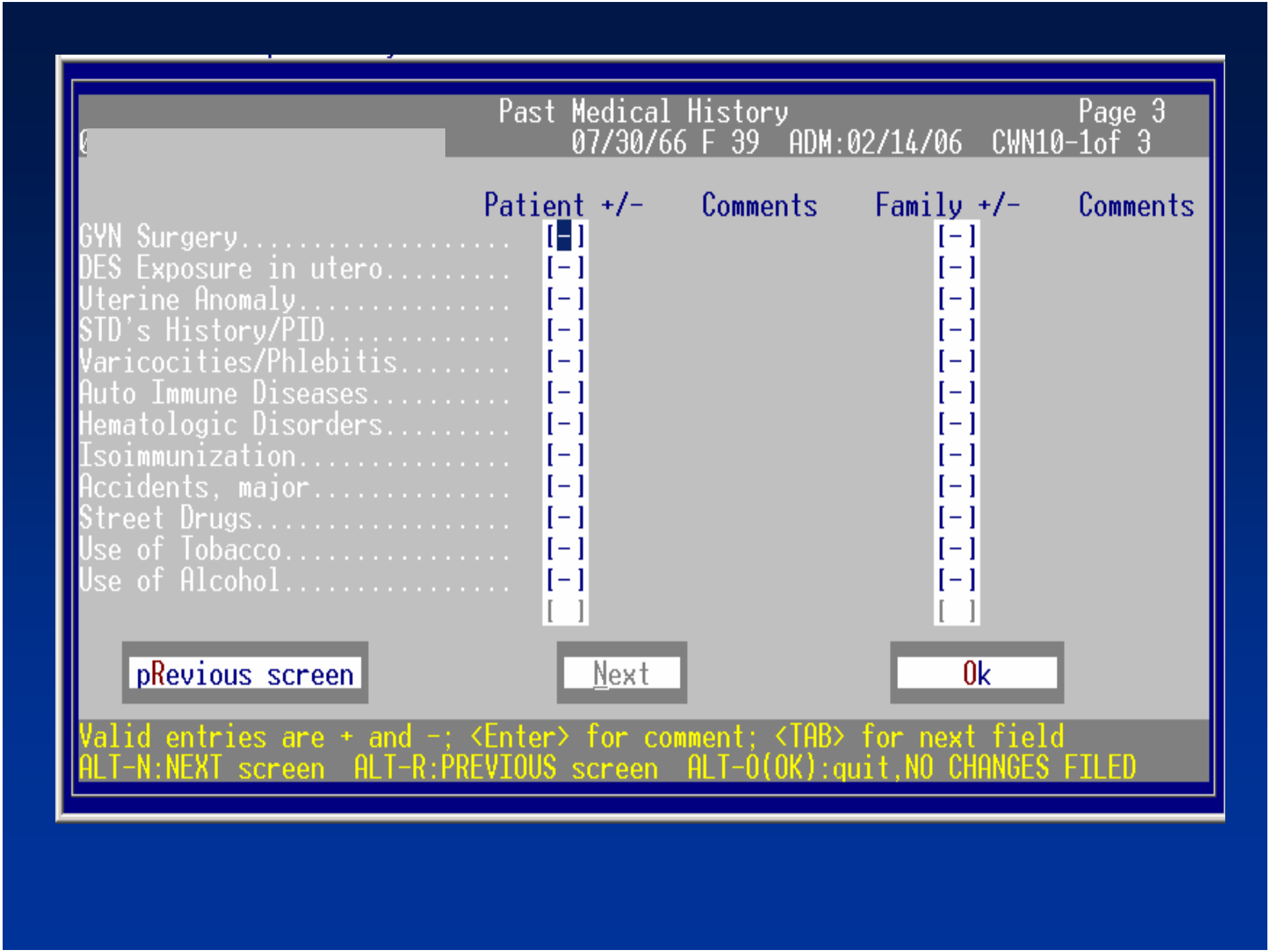
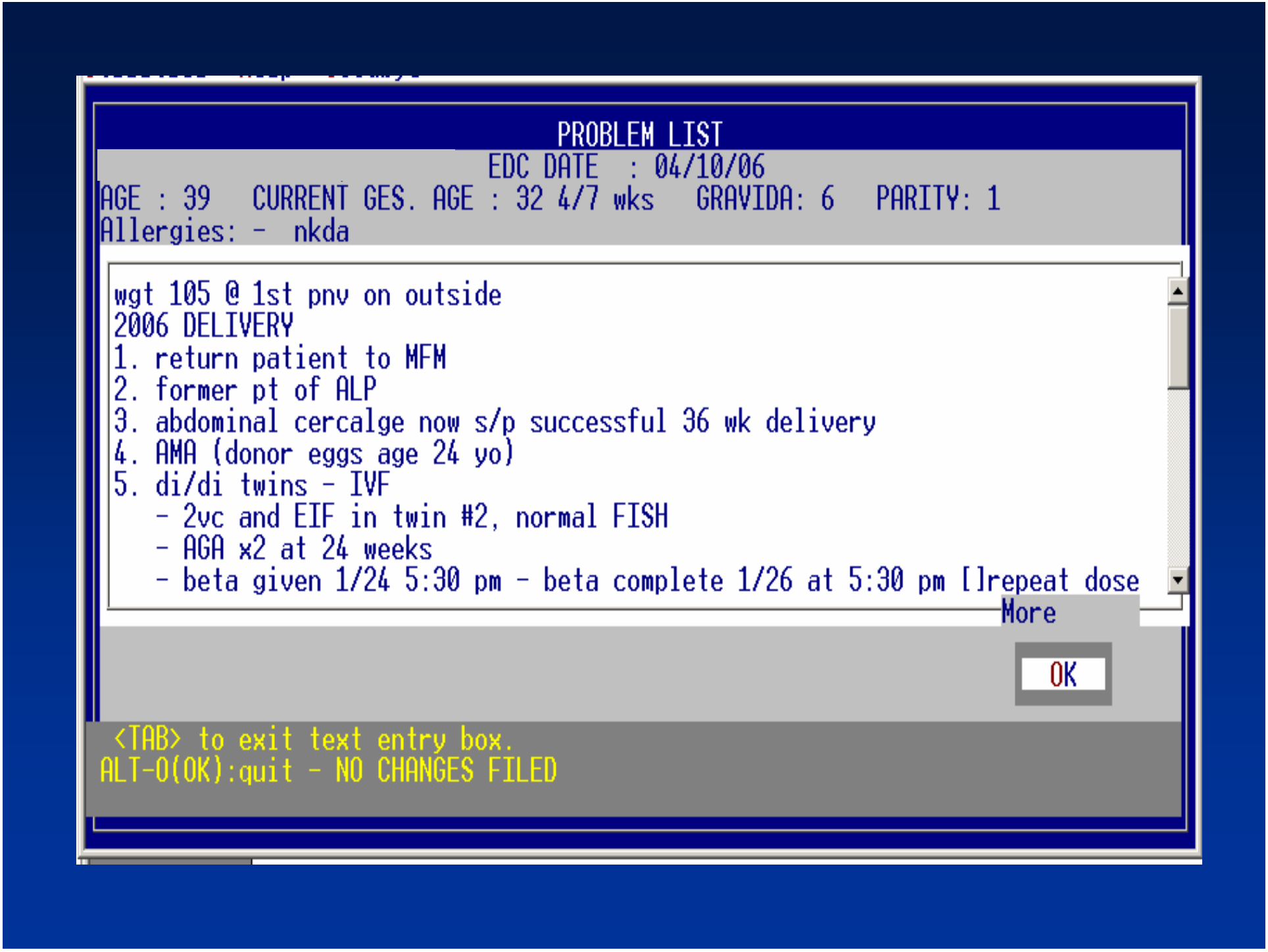
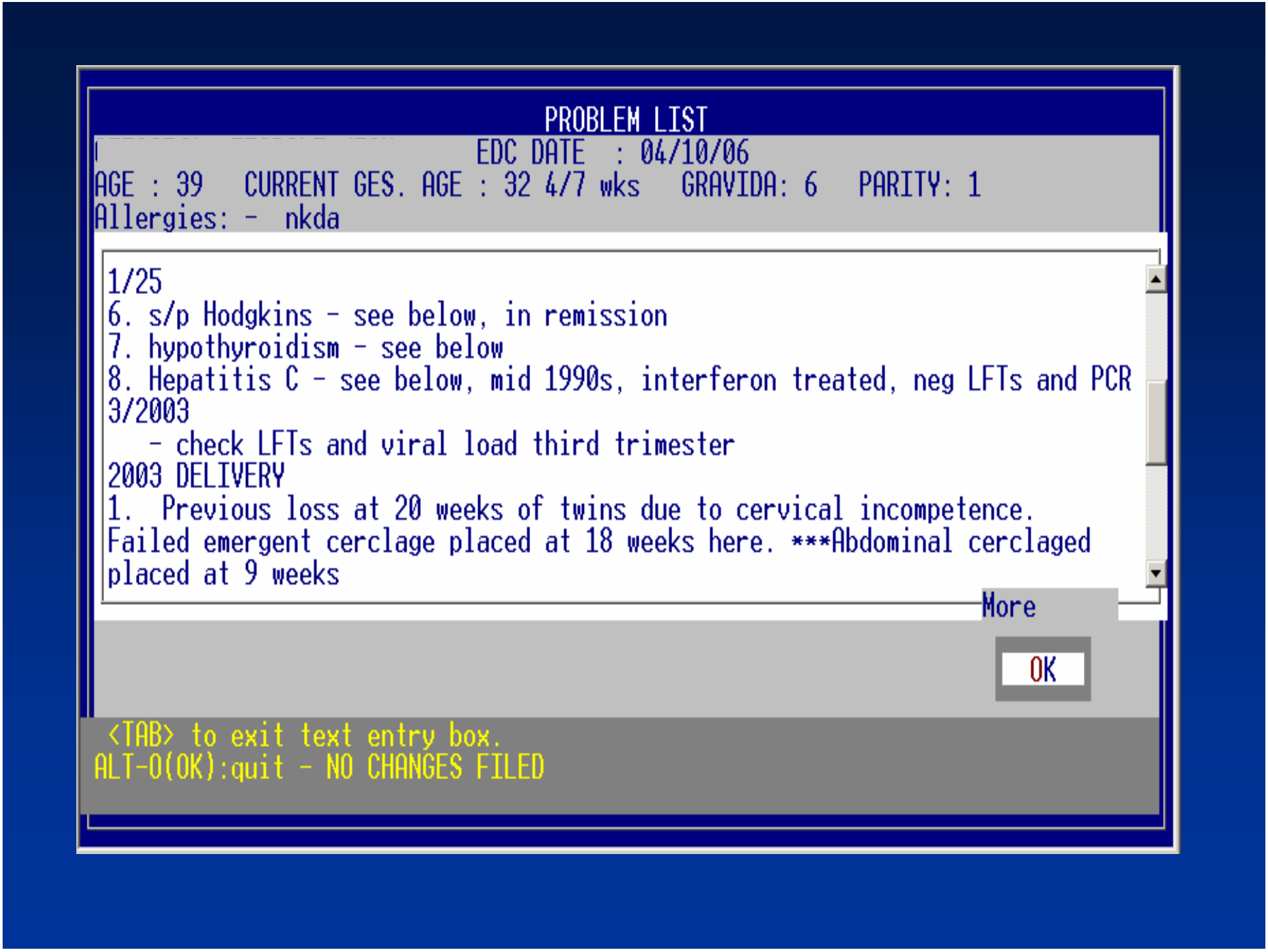

Genetic Screening
Page 1
11/26/70 F 35 ADM:02/12/06 - 02/14/06 EDC
Questions applicable to patient, FOB, either's family
+/-
Comments
Patient age 35 or older [+]
Down syndrome [-]
Neural Tube Defect [-]
Hemophilia [-]
Muscular dystrophy [-]
Cystic Fibrosis [-]
Huntington's chorea [-]
Sickle cell (Dx or Trait) [-]
Mental retardation [-]
Jewish,French-Canadian,Italian,Mediterranean,Asian [-]
Meds/Drugs since LMP [-]
Pt. or FOB has child w/ birth defect not listed above [-]
Other [-]
Ok

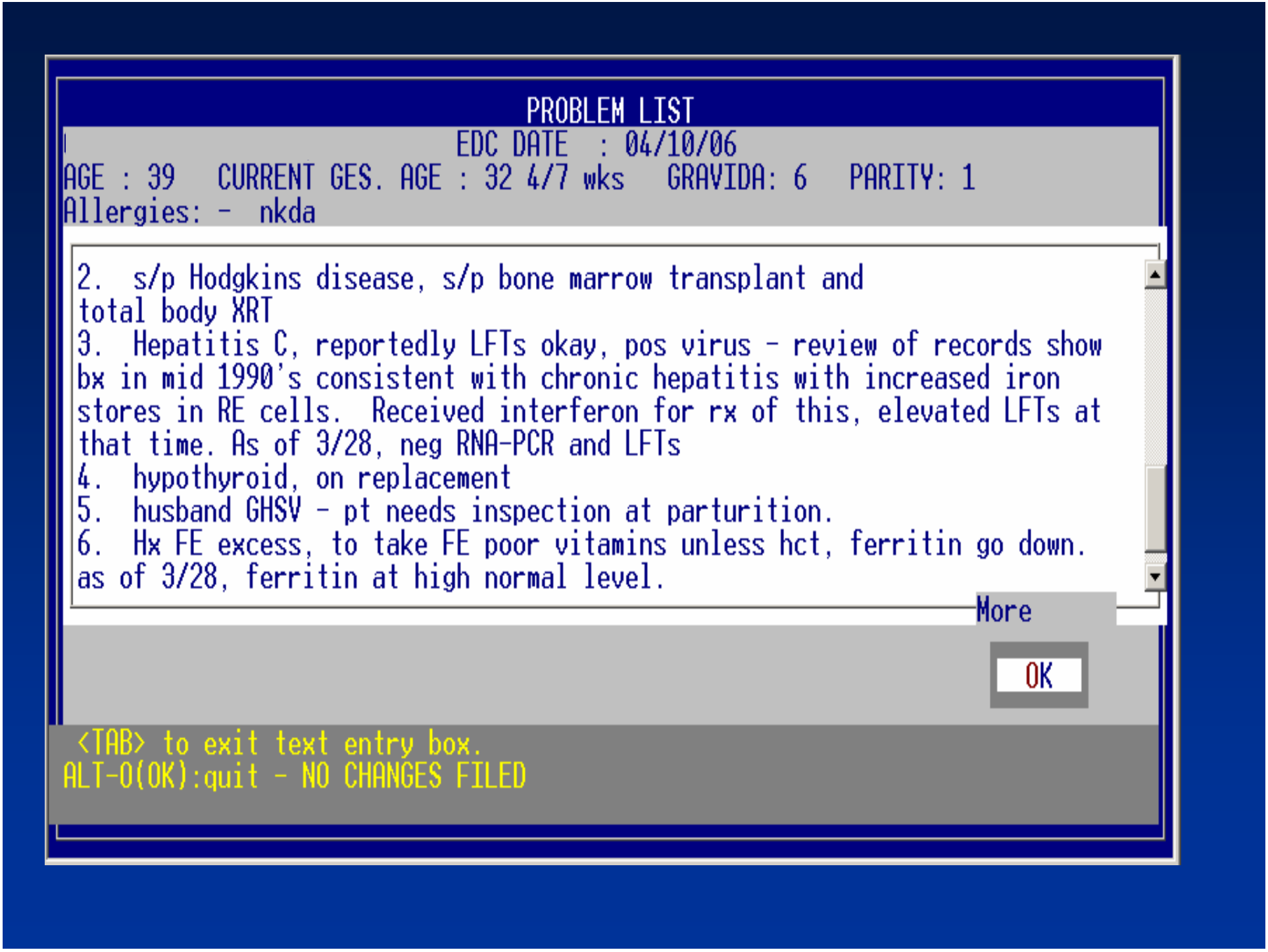

Past Medical History
Page 1
07/30/66 F 39 ADM:02/14/06 CWN10-1of 3
Patient +/- Comments Family +/- Comments
Thyroid/Endocrine Function.... [+] HYPOTHYROID RX WITH [-]
Hepatitis/Liver Disease..... [+] H/O HEP C LFT'S NEA [-]
Infertility..... [+] IVF SEEN BY DR GINS [-]
Cancer..... [+] HODGKINS DISEASE BON [-]
Blood Tranfusion Hx..... [+] SEVERAL RELATED TO H [-]
Operations/Hospitalizations... [+] 1988-BONE MARROW TRA [-]
Chicken Pox History..... [+] in childhood [ ]
Domestic Violence..... [ ] pt denies [-]
Allergies..... [-] nkda [-]
Headaches..... [-] tylenol prn [-]
Abnormal PAP hx..... [-] LAST PAP < 1YR [-]
Herpes..... [-] ? HUSBAND HAS OUTBRE [-]
Anesthetic Complications..... [-] denies [-]
pRevious screen
Next
Ok
Valid entries are + and -; <Enter> for comment; <TAB> for next field
ALT-N:NEXT screen ALT-R:PREVIOUS screen ALT-O(OK):quit,NO CHANGES FILED


Past Medical History
Page 2
07/30/66 F 39 ADM:02/14/06 CWN10-1of 3
Patient +/- Comments
Family +/- Comments
HIV Risk [-] TESTED NEG @ CRM [-]
Other [-] pt denies depression [ ]
Neurological [-] [-]
Seizures [-] [-]
Diabetes [-] [+] SISTER TYPE 1
Pulmonary Disease [-] [-]
Asthma [-] [-]
Tuberculosis [-] [-]
Cardiac Disease/RHF [-] [-]
Mitral Valve Prolapse [-] [-]
GI Disorders [-] [-]
Kidney Disease/UTI [-] [-]
Hypertension [-] [-]
pRevious screen
Next
Ok
Valid entries are + and -; <Enter> for comment; <TAB> for next field
ALT-N:NEXT screen ALT-R:PREVIOUS screen ALT-O(OK):quit,NO CHANGES FILED


Past Medical History
Page 3
07/30/66 F 39 ADM:02/14/06 CWN10-1of 3
Patient +/-
Comments
Family +/-
Comments
GYN Surgery.....
DES Exposure in utero.....
Uterine Anomaly.....
STD's History/PID.....
Varicocities/Phlebitis.....
Auto Immune Diseases.....
Hematologic Disorders.....
Isoimmunization.....
Accidents, major.....
Street Drugs.....
Use of Tobacco.....
Use of Alcohol.....
pRevious screen
Next
Ok
Valid entries are + and -; <Enter> for comment; <TAB> for next field
ALT-N:NEXT screen ALT-R:PREVIOUS screen ALT-O(OK):quit,NO CHANGES FILED


PROBLEM LIST
EDC DATE : 04/10/06
AGE : 39 CURRENT GES. AGE : 32 4/7 wks GRAVIDA: 6 PARITY: 1
Allergies: - nkda
wgt 105 @ 1st pnv on outside
2006 DELIVERY
1. return patient to MFM
2. former pt of ALP
3. abdominal cercalge now s/p successful 36 wk delivery
4. AMA (donor eggs age 24 yo)
5. di/di twins - IVF
- 2vc and EIF in twin #2, normal FISH
- AGA x2 at 24 weeks
- beta given 1/24 5:30 pm - beta complete 1/26 at 5:30 pm []repeat dose
More
OK
<TAB> to exit text entry box.
ALT-O(OK):quit - NO CHANGES FILED


PROBLEM LIST
EDC DATE : 04/10/06
AGE : 39 CURRENT GES. AGE : 32 4/7 wks GRAVIDA: 6 PARITY: 1
Allergies: - nkda
1/25
6. s/p Hodgkins - see below, in remission
7. hypothyroidism - see below
8. Hepatitis C - see below, mid 1990s, interferon treated, neg LFTs and PCR
3/2003
- check LFTs and viral load third trimester
2003 DELIVERY
1. Previous loss at 20 weeks of twins due to cervical incompetence.
Failed emergent cerclage placed at 18 weeks here. ***Abdominal cerclaged
placed at 9 weeks
More
OK
<TAB> to exit text entry box.
ALT-O(OK):quit - NO CHANGES FILED


PROBLEM LIST
EDC DATE : 04/10/06
AGE : 39 CURRENT GES. AGE : 32 4/7 wks GRAVIDA: 6 PARITY: 1
Allergies: - nkda
2. s/p Hodgkins disease, s/p bone marrow transplant and
total body XRT
3. Hepatitis C, reportedly LFTs okay, pos virus - review of records show
bx in mid 1990's consistent with chronic hepatitis with increased iron
stores in RE cells. Received interferon for rx of this, elevated LFTs at
that time. As of 3/28, neg RNA-PCR and LFTs
4. hypothyroid, on replacement
5. husband GHSV - pt needs inspection at parturition.
6. Hx FE excess, to take FE poor vitamins unless hct, ferritin go down.
as of 3/28, ferritin at high normal level.
More
OK
<TAB> to exit text entry box.
ALT-O(OK):quit - NO CHANGES FILED

PROBLEM LIST

EDC DATE : 04/10/06

AGE : 39 CURRENT GES. AGE : 32 4/7 wks GRAVIDA: 6 PARITY: 1

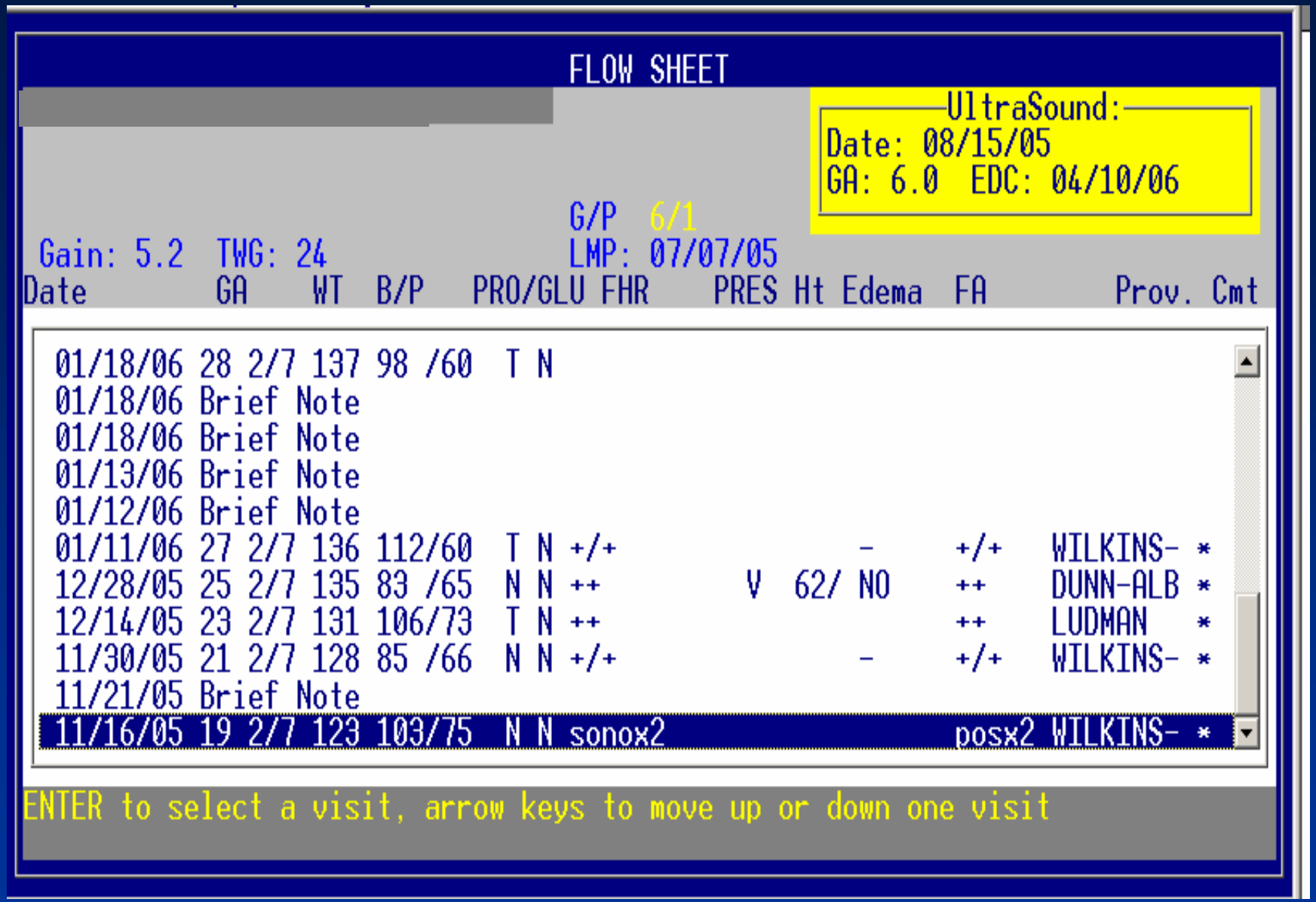
Allergies: - nkda

total body XRT
3. Hepatitis C, reportedly LFTs okay, pos virus - review of records show bx in mid 1990's consistent with chronic hepatitis with increased iron stores in RE cells. Received interferon for rx of this, elevated LFTs at that time. As of 3/28, neg RNA-PCR and LFTs
4. hypothyroid, on replacement
5. husband GHSV - pt needs inspection at parturition.
6. Hx FE excess, to take FE poor vitamins unless hct, ferritin go down. as of 3/28, ferritin at high normal level.
7. AMA: declines afp and amnio. See notes.

More

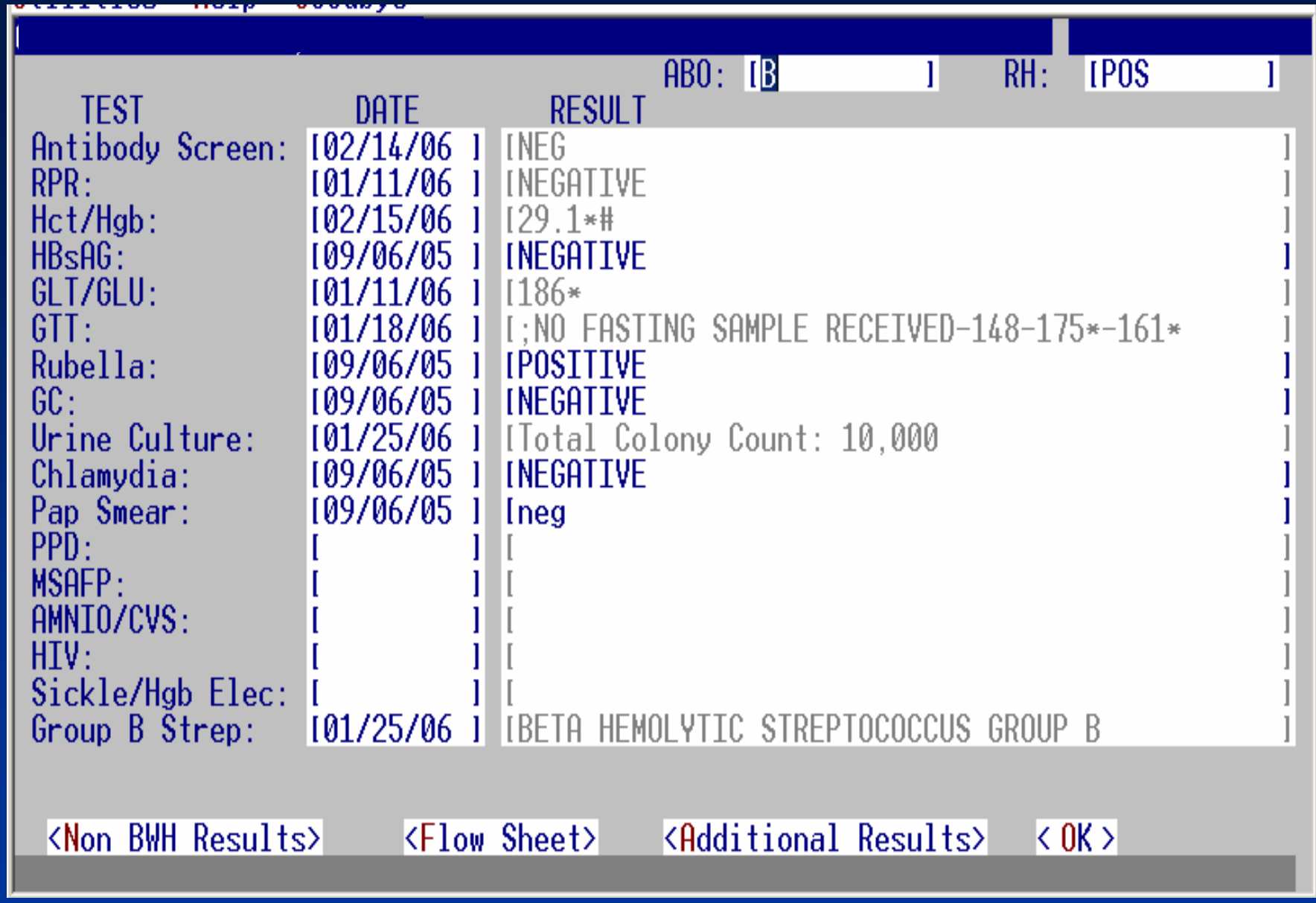
OK

<TAB> to exit text entry box.
ALT-O(OK):quit - NO CHANGES FILED


FLOW SHEET
UltraSound:
Date: 08/15/05
GA: 6.0 EDC: 04/10/06
G/P 6/1
Gain: 5.2 TWG: 24
LMP: 07/07/05
Date GA WT B/P PRO/GLU FHR PRES Ht Edema FA Prov. Cmt
01/18/06 28 2/7 137 98 /60 T N
01/18/06 Brief Note
01/18/06 Brief Note
01/13/06 Brief Note
01/12/06 Brief Note
01/11/06 27 2/7 136 112/60 T N +/+ - +/+ WILKINS- *
12/28/05 25 2/7 135 83 /65 N N ++ V 62/ NO ++ DUNN-ALB *
12/14/05 23 2/7 131 106/73 T N ++ ++ LUDMAN *
11/30/05 21 2/7 128 85 /66 N N +/+ - +/+ WILKINS- *
11/21/05 Brief Note
11/16/05 19 2/7 123 103/75 N N sonox2 posx2 WILKINS- *
ENTER to select a visit, arrow keys to move up or down one visit

Note

- first US here - 2v cord and EIF - did not get NL scans done to her knowledge,
- discussed increased risk for trisomy 21 with EIF, also with second finding of 2v cord; cardiac and renal are normal; given 24 yo egg donor even increasing her risk 3-5 fold would still result in an absoulte risk which is small (majority would be normal) - risk of 1/200 for amnio discussed,
- selective reduction discussed - will talk to husband and let me know


ABO: [B]
RH: [POS]
TEST
DATE
RESULT
Antibody Screen: [02/14/06] [NEG]
RPR: [01/11/06] [NEGATIVE]
Hct/Hgb: [02/15/06] [29.1*#]
HBsAG: [09/06/05] [NEGATIVE]
GLT/GLU: [01/11/06] [186*]
GTT: [01/18/06] [;NO FASTING SAMPLE RECEIVED-148-175*-161*]
Rubella: [09/06/05] [POSITIVE]
GC: [09/06/05] [NEGATIVE]
Urine Culture: [01/25/06] [Total Colony Count: 10,000]
Chlamydia: [09/06/05] [NEGATIVE]
Pap Smear: [09/06/05] [neg]
PPD: [ ] [ ]
MSAFP: [ ] [ ]
AMNIO/CVS: [ ] [ ]
HIV: [ ] [ ]
Sickle/Hgb Elec: [ ] [ ]
Group B Strep: [01/25/06] [BETA HEMOLYTIC STREPTOCOCCUS GROUP B]
<Non BWH Results>
<Flow Sheet>
<Additional Results>
< OK >

INPATIENT ADMISSION SHEET

NEWBORN FACESHEET

BRIGHAM AND WOMEN'S HOSPITAL
BOSTON, MA 02115

ADMIT DATE: 02/16/06
ADMIT TIME: 22:46

MATERNAL DATA

MOTHER'S MR#: 20504163
MOTHER'S ROOM #: LAB-15

Age: 24 G: P: EDC: 03/01/06 GA: 38 wks.
Blood Type: A+ Antibody screen: NEG
Prenatal Information:

RUBELLA	Positive
RPR	
HBsAg	Negative
GC	
Chlamydia	
GBS	Negative
TOX scrn	
PPD	

LABOR
Room: L04 Length of ROM: 7 hrs. 31 min.
Complications of labor:
None
Birth Weight: 5 LBS. 2 OZ.
(2325 GRAMS)

DELIVERY
Date: 02/16/06 Time: 22:46
Route: Vaginal
Spontaneous
Apgar Score: 9 9
Length: 18 INCHES
(45 CMS.)

SEX: F BIRTHDATE: 02/16/06 AGE: 0

ADMIT DIAGNOSIS: NEWBORN

ADMIT DIAGNOSIS CODE: V30.00 ADMIT STATUS: NEWBORN

SERVICE: NYC

BRIGHAM AND WOMEN'S HOSPITAL
A Teaching Affiliate of Harvard Medical School
75 Francis Street, Boston, Massachusetts 02115

NEWBORN SUMMARY

Infant's Last Name ______

ANTEPARTUM

MATERNAL DATA
Age 24 G 3 P 2 EDC 3-1-06 Blood Type A+

Prenatal Care Utilization:
☐ Visits <5
☐ Initial Visit > 28 wks.

PRENATAL HISTORY For Prenatal Screens - See infant face sheet
G3P2 EDC 3/1/06 @38¹
GBS (–)
Hep (–)
RI
Present pregnancy → 2VC, TOF, declined Amnio

INTRAPARTUM

LABOR OB Provider ______
ROM ______ hrs 2/16/06 @ 3¹⁵pm.

DELIVERY
Date 2/16/06 Time 10⁴⁶pm

Sepsis Risk Factors (check all that apply):
☒ None
☐ Gp B Strep +
☐ Fever ____ max. temp.
☐ Other ______

Route:
☒ Vaginal
☐ C/Sect (reason) ______
☐ Meconium → ☐ Intubated → ☐ Mec. ↓ cord ☐ yes ☐ no

HC 34cm
Len 45cm

Delivery Room Course:
Called to L+D d/t prenatal Dx of TOF, baby born thru clear fluid, spontaneous cry at table. Brought to warmer dried, suctioned, stimulated. AFOS, palate in tact, Ø neck masses, clavicles in tact, good aeration B/L, RRR, ØM appreciated (crying baby), 2+ fem pulses, abd ND, 2 vessel cord, nl ♀ ext genitalia, anus appears patent, Ø dimple, 5 digits x4 ext, Ø [illegible] were noted

Apgars 9 (1) 9 (5) ___ (10)
Sex: ☐ M ☒ F

BW 2330 kg
5.2 lbs

ASSESSMENT To NICU Triage for further eval RN/MD D N 8 G 4

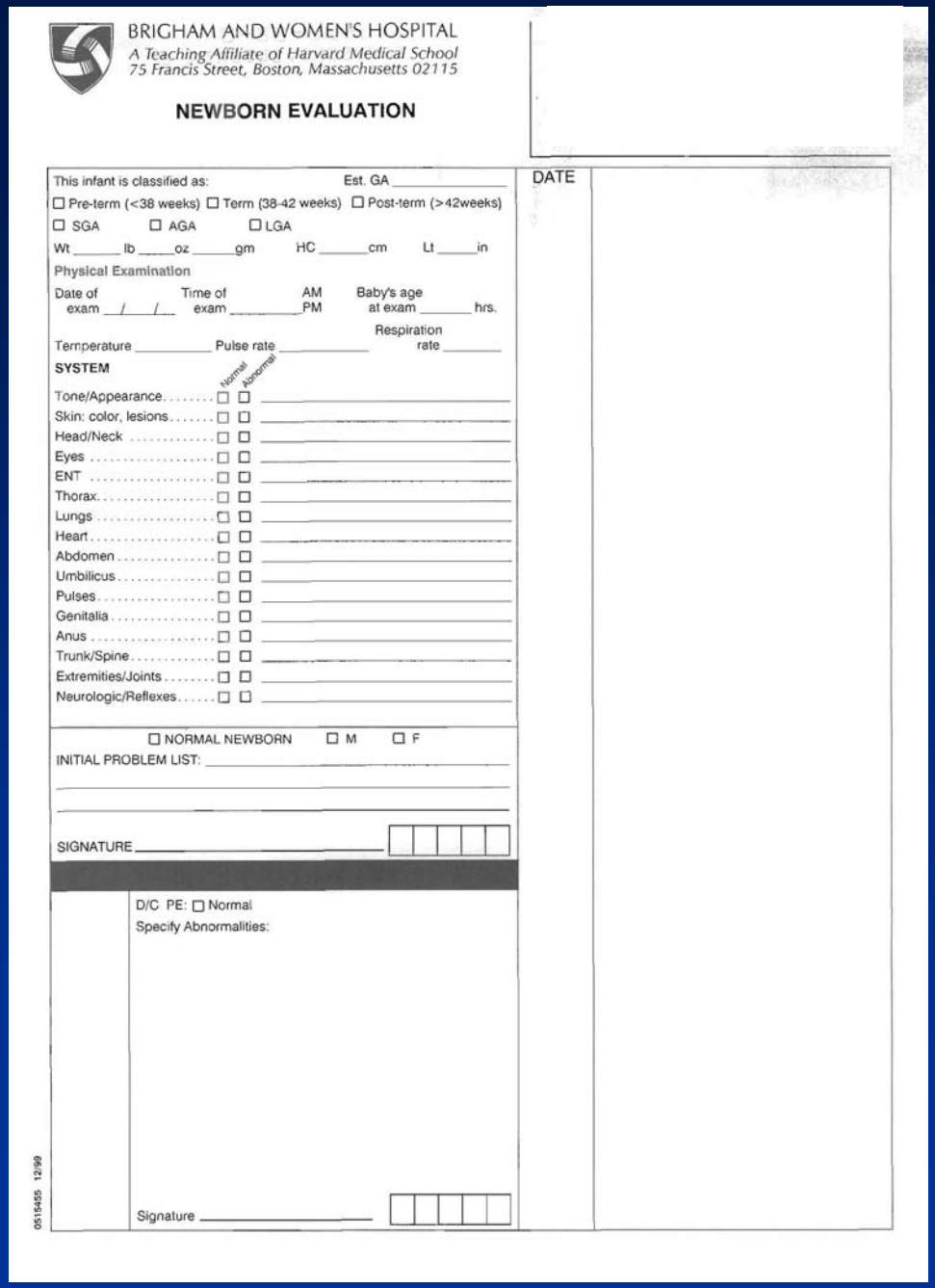
NEONATAL

Feeding Plans: ☐ breast ☐ bottle
D/C date: __/__/__
Bilirubin: Max Bili ______ Date ______ ☐ Photo Rx
Diagnoses: ☐ Normal Newborn
Blood Type: ______ Coombs: ______
Hep B Imm. ☐ Given ☐ Not Given
Newborn Screen (date) ______
Age (Hrs.) ______
D/C Wt. ____ lb ____ oz ____ gm
Feeding Method at d/c: ☐ breast ☐ bottle

FOLLOW UP

Provider/Clinic: ______ City: ______
Appointment: ☐ 2 weeks (advised to call) ☐ Date __/__/__ Home Tel # ______
Referrals: ☐ VNA ☐ DSS Medications: ______
Plans/Instructions: ______

Questions can be directed to Newborn Medicine, (617) 732-7739 Signature ______



BRIGHAM AND WOMEN'S HOSPITAL
A Teaching Affiliate of Harvard Medical School
75 Francis Street, Boston, Massachusetts 02115

NEWBORN EVALUATION

DATE

This infant is classified as: Est. GA _____
☐ Pre-term (<38 weeks) ☐ Term (38-42 weeks) ☐ Post-term (>42weeks)
☐ SGA ☐ AGA ☐ LGA
Wt _____ lb ____oz _____gm HC _____cm Lt ____in

Physical Examination

Date of exam __/__/__ Time of exam _____ AM PM Baby's age at exam _____ hrs.

Temperature _____ Pulse rate _____ Respiration rate _____

SYSTEM	Normal	Abnormal	
Tone/Appearance	☐	☐	
Skin: color, lesions	☐	☐	
Head/Neck	☐	☐	
Eyes	☐	☐	
ENT	☐	☐	
Thorax	☐	☐	
Lungs	☐	☐	
Heart	☐	☐	
Abdomen	☐	☐	
Umbilicus	☐	☐	
Pulses	☐	☐	
Genitalia	☐	☐	
Anus	☐	☐	
Trunk/Spine	☐	☐	
Extremities/Joints	☐	☐	
Neurologic/Reflexes	☐	☐	

☐ NORMAL NEWBORN ☐ M ☐ F

INITIAL PROBLEM LIST: _____

SIGNATURE _____

D/C PE: ☐ Normal
Specify Abnormalities:

Signature _____

0515455 12/99

Conclusion

- Tool should be developed for transmission of OB/perinatal history as foundation for pediatric tool.
- Prenatal and perinatal data may be important to predicting pediatric disease.