Newborn Screening Program

PerkinElmer®
For the Better

Nemours®
Alfred I. duPont Hospital for Children

Who We Are – Nemours Team

- **Your contact point for:**
  - Abnormal Results Notification
  - Education
  - Genetic Support
- **Monday to Friday 8 a.m. to 4:30 p.m.**
- **(302) 651-5079**
  denewbornscreening@nemours.org
- **After Hours**
  - (302) 651-4000
  - Ask for the Diagnostic Referral Physician on call

Nemours Team

- Michael Cellucci, MD, *Medical Director*
- Kathryn Tullis, PhD, *Program Manager*
- Jessica Carmona, *Program Coordinator*
- Deborah Consolini, MD, *Medical Consultant*
- Matthew Demczko, MD, *Medical Consultant*
- Michael Fox, MD, *Medical Consultant*
- Jeffrey Malatack, MD, *Medical Consultant*
- Christopher Raab, MD, *Medical Consultant*
- Catherine Soprano, MD, *Medical Consultant*

Client Service Team - PerkinElmer

- **Single point of contact for all submitters at (866) 463-6436**
  - Monday – Friday 8 a.m. to 5 p.m. Eastern Standard Time
- **Contact CSR's for non clinical issues such as:**
  - Reordering supplies (filter papers, brochures, UPS Supplies);
  - Requests for Internet Access ID's; Password Resets; Account Reactivation;
  - Normal Results;
  - UPS Tracking for specimens through UPS Quantum View
  - Triage calls to appropriate resource for resolution;
  - General requests i.e., specimen requirements, confirmation of sample receipt, CPT Codes, CAP & CLIA Certifications
- **Client Service Team will contact Submitters for the following:**
  - Collecting Missing Demographics from Submitters – Request for Information Sheets are Faxed Daily
    - *Birth Date & Time*
    - *Collection Date & Time*
  - Notification of Unacceptable Specimens to Request Repeat
  - Notification of Less Than 24 Hour Specimens
  - Corrections to Reports must be in writing and faxed to PerkinElmer Genetics (412) 220-0785 or 0784
  - Obtaining PCP information

Who We Are – PerkinElmer's Client Services Team

- Monday to Friday
  8 a.m. to 5 p.m.
  (US Eastern Time)
- (866) 463-6436

Specimen Collection Requirements

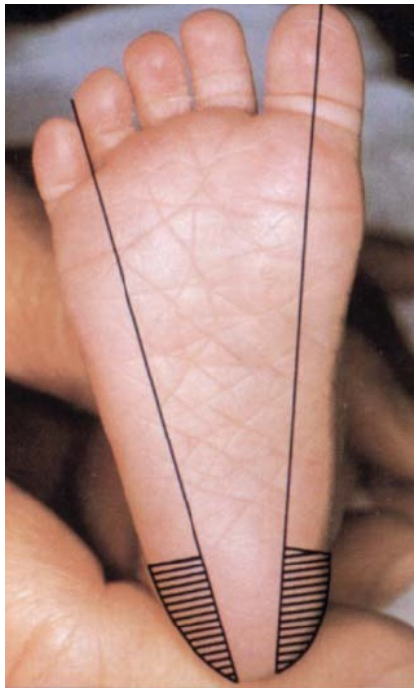
- **Blood Collection Form**
  - Always, collect blood specimens utilizing filter papers approved by the DE DOH.
  - Healthcare Provider must ensure that submitted specimens are legible and completely filled out with demographic information including PCP information.
  - Needed to ensure proper interpretation and reporting of results.
- **Timing**
  - Collect between 24-48 hours of age, as close to 48 hours as possible.
  - If discharged before 24 hours of age:
    - *collect at discharge*
    - *collect a repeat specimen at 48 hours of age*
  - Critically ill infants should be screened by 48 hours of age.

Specimen Collection Requirements

- **If transfused**
  - A pre-transfusion specimen is essential
  - If not possible;
    - *Draw one specimen at 24-48 hours of age*
    - *Draw a repeat specimen 3 days after the last transfusion*
- **Screen can be done on patients of any age, except:**
  - Cystic Fibrosis which is not valid after 3 months of age

Specimen Collection Technique

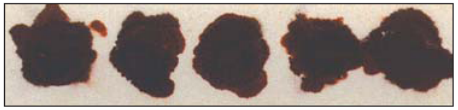
- Fill each of the 5 circles on the filter paper;
- One, free-flowing drop of blood;
- Do not layer more than one drop;
- On one side only;
- Make sure the blood has saturated through.



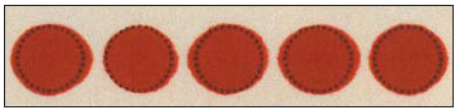
Specimen Handling

- **Air dry**
  - Flat surface
  - Away from heat and light
  - 3-4 hours
- **Do not**
  - Stack
  - Allow the blood spots to touch other surfaces when drying
- **Ship promptly**
- **If unable to ship immediately**
  - Refrigerate, or
  - Store in a plastic bag in a freezer

1. Specimen quantity insufficient for testing.

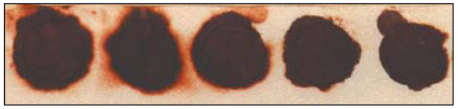


2. Specimen appears scratched or abraded.



3. Specimen not dry before mailing.

4. Specimen appears supersaturated.



5. Specimen appears diluted, discolored or contaminated.

6. Specimen exhibits serum rings.

7. Specimen appears clotted or layered.

Necessary Demographics:

Demographic	Purpose	Consequence of Missing or Inaccurate Information
Baby's Last Name	Identification	May cause a hindrance in the prompt notification of results and possible misidentification of newborn.
Draw Date	To ensure sample is drawn after 24 hours of age	Certain disorders may not be detectable before 24 hours of age. Ideal period for specimen collection is 24 to 48 hours, collection after 48 hours of age is permissible. It is very important that the date and time of collection is recorded so that the results can be properly interpreted. **If draw date is before 24 hours**, sample is identified as **"less than 24 hours of age"** and PerkinElmer Genetics will Request a Repeat Specimen at no charge to submitter. The Draw Date will also identify if a specimen was received at PerkinElmer Genetics **30 days after draw date**. If yes, the specimen would be identified as **"Unacceptable"** and PerkinElmer Genetics will Request a Repeat Specimen at no charge to the submitter.
Draw Time	To ensure sample is drawn after 24 hours of age	As above (Draw Date)

Important!

Necessary Demographics: (Continued)

Demographic	Purpose	Consequence of Missing or Inaccurate Information
Mother's Name & Telephone	Identification and correct reporting of results	This information is most helpful to the pediatrician to identify correct baby as baby names are often not the same as mother.
Hospital of Birth & Sex	Identification and correct reporting of results	May cause a hindrance in the prompt notification of results and possible misidentification of newborn.
Birth Date	Identification and determination of > 24 hour specimen Important!	There are certain disorders that may not be detectable before 24 hours of age. The best time period for specimen collection is 24 to 48 hours, however collection after 48 hours of age is permissible. It is very important that the date and time of collection is recorded on the filter paper form so that the results can be properly interpreted. **If draw date is before 24 hours**, sample is identified as **"less than 24 hours of age"** and PerkinElmer Genetics will Request a Repeat Specimen at no charge to submitter.
Birth Time	A determining factor to identify if the specimen was drawn before 24 hours of age. Important!	There are certain disorders that may not be detectable before 24 hours of age. The best time period for specimen collection is 24 to 48 hours, however collection after 48 hours of age is permissible. It is very important that the date and time of collection is recorded on the filter paper form so that the results can be properly interpreted. **If draw date is before 24 hours**, sample is identified as **"less than 24 hours of age"** and PerkinElmer Genetics will Request a Repeat Specimen at no charge to submitter.

Necessary Demographics: (Continued)

Demographic	Purpose	Consequence of Missing or Inaccurate Information
Gestation (weeks) Birth Weight (grams) Multiple Births	Correct reporting of results.	As premature babies, low birth weight babies, and multiples have high probability of mild elevations in parts of the screening (due to the prematurity, not metabolic disease). This information helps us in reviewing results and reporting correctly given the baby's clinical condition. Normal ranges for CAH screening vary SIGNIFICANTLY depending on birth weight.
Transfusion	Transfusions may cause missed diagnosis in newborns, therefore, a pre-transfusion specimen is essential. If this is not possible obtain one specimen at 24-48 hours of age and a repeat specimen 3 days after the last transfusion.	If a transfusion is necessary, a specimen should be collected before the transfusion regardless of the age of the baby. While transfusions cause problems because of the dilution effect and the time it takes for various analytes to reach their concentrations; the following tests can be performed 72 hours after the transfusion: Amino Acid Disorders, Acylcarnitine Disorders, Galactosemia, Hypothyroidism (TSH & T4), Congenital Adrenal Hyperplasia, Biotinidase Deficiency and Cystic Fibrosis.
Physician's Address & Telephone	Correct reporting of results. Important!	May delay the notification of results to physician and delay medical intervention.
Submitter	Identifies facility for notification of results, type of testing based on submitter profile in our LIS system, delivery of reports and invoicing.	Misdirection of results, reports and invoices.
Address if other than birth facility	Correct reporting of results.	May cause a hindrance in the prompt notification of results to physician of record.

Specimen Rejection Criteria – Unacceptable Specimens

- No blood or quantity not sufficient for testing
- Specimen oversaturated
- Specimen diluted, discolored or contaminated
- Specimen exhibits serum rings
- Specimens received > 30 days after collection date
- Blood not soaked through
- Clots on surface of blood spot
- Blood on both sides
- Mailed while blood is wet
- Specimen got wet in transit
- Surface of specimen abraded
- Specimen double-spotted

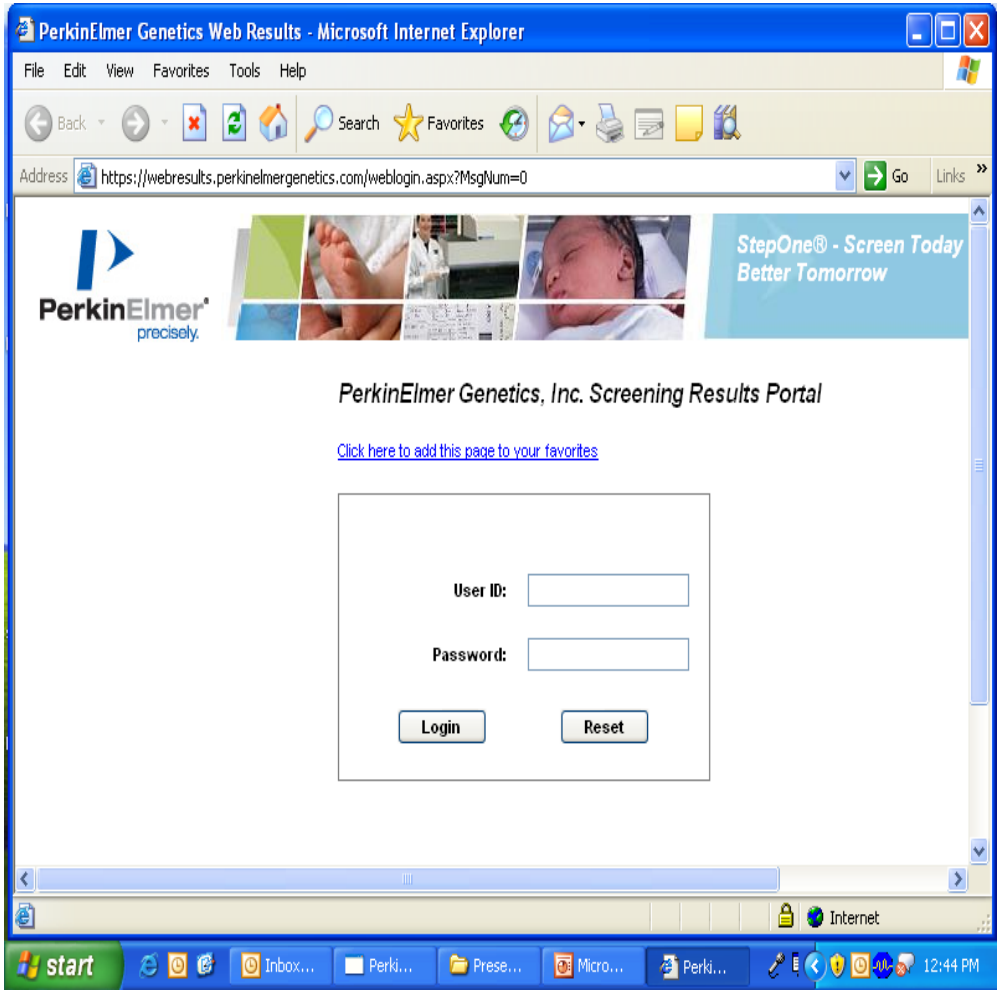
Results Portal – Secure Access to Results

- On the PerkinElmer Genetics website
- Secure through Secure Socket Layer (SSL)
- 128 bit encryption
- Search for individual results
- Download a series of results
- In Adobe Acrobat format

Obtaining ID & Password

- First, a Submitter Number must be assigned
- Each individual requiring access to on-line reports must complete an Internet Access Request Form
- Fax completed form to our IT Department at (412) 220-0784
- ID & Password will be faxed to user on an instruction sheet
- Only specimens submitted by your facility will be accessible through your ID

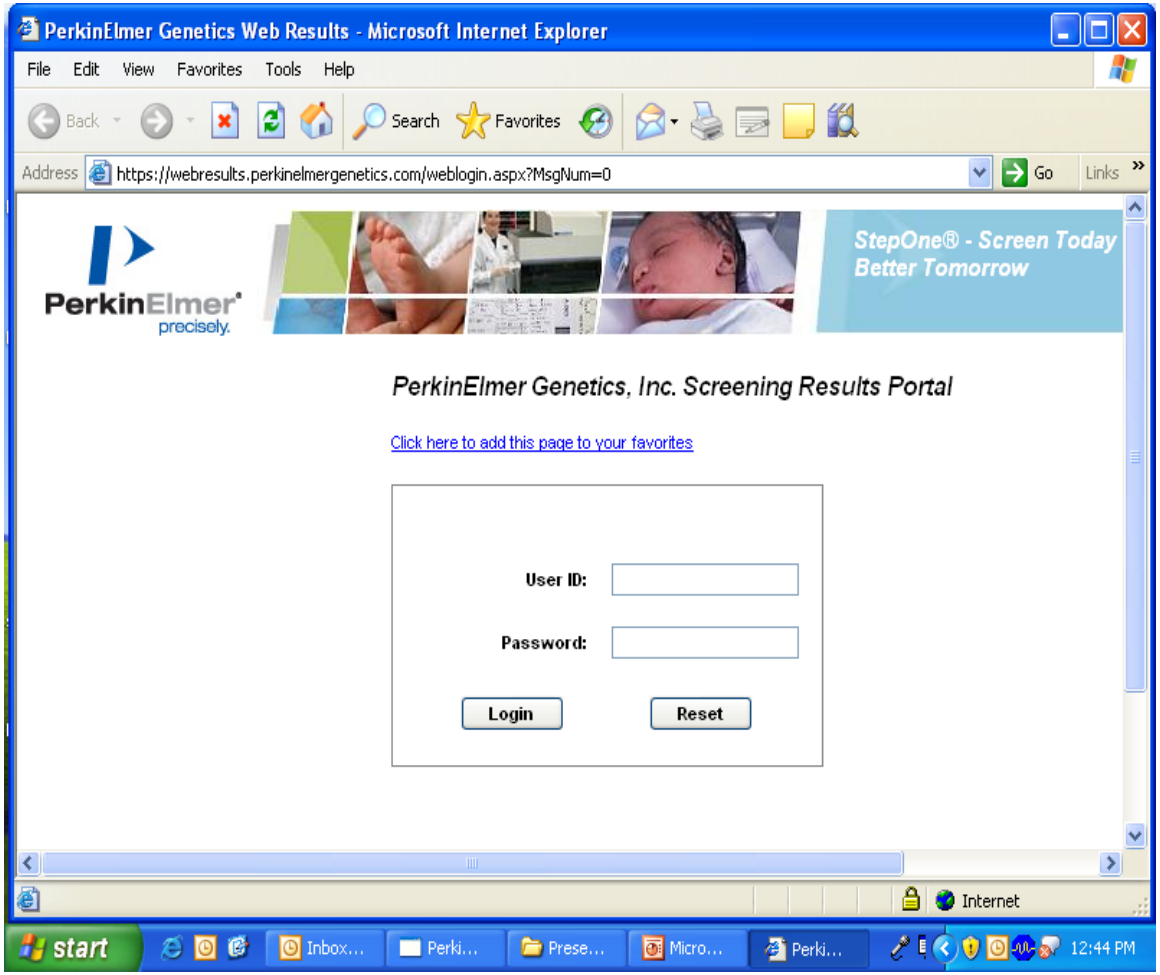
Results Portal – Web Site



- Go to:
  http://www.perkinelmergenetics.com
  or
  https://resultsportal.perkinelmergenetics.com

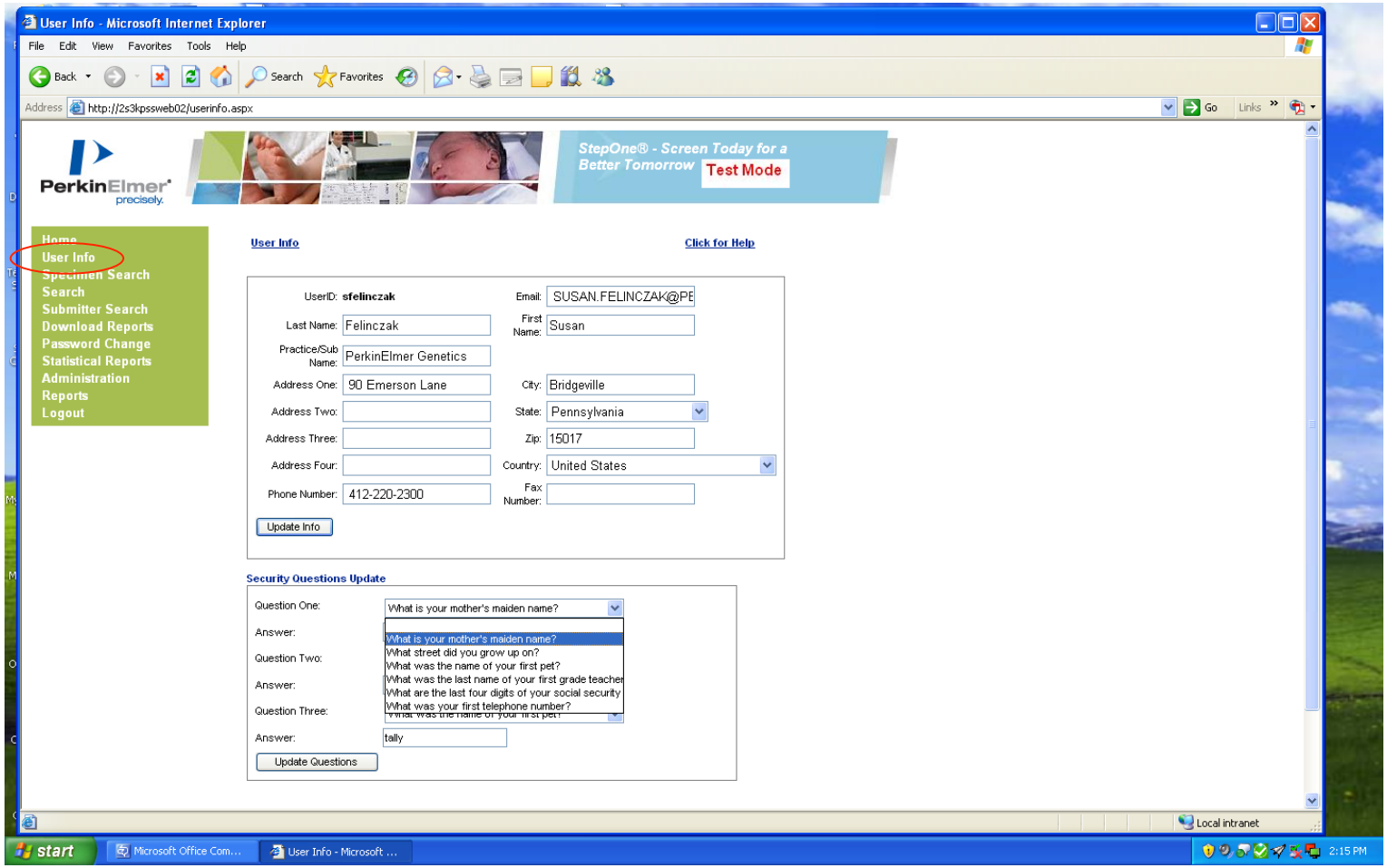
- Click on **"login"** in the **"Results Center"** section.

Results Portal – Login

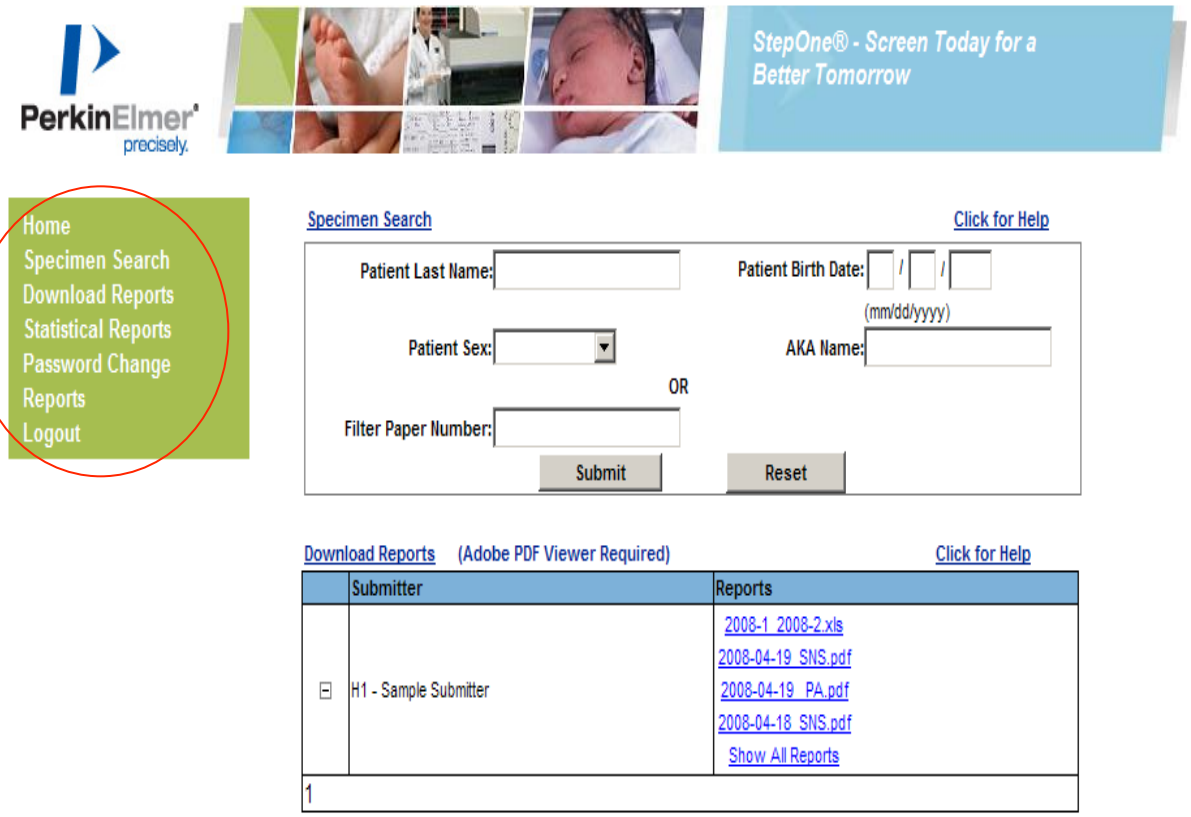


- Enter **username(1)** and **password(2)** and then click **"Login"**.
- The yellow lock signifies you are in a 128 bit Secure Socket Layer Session (SSL).
- Account User ID and Password are case sensitive.

Security

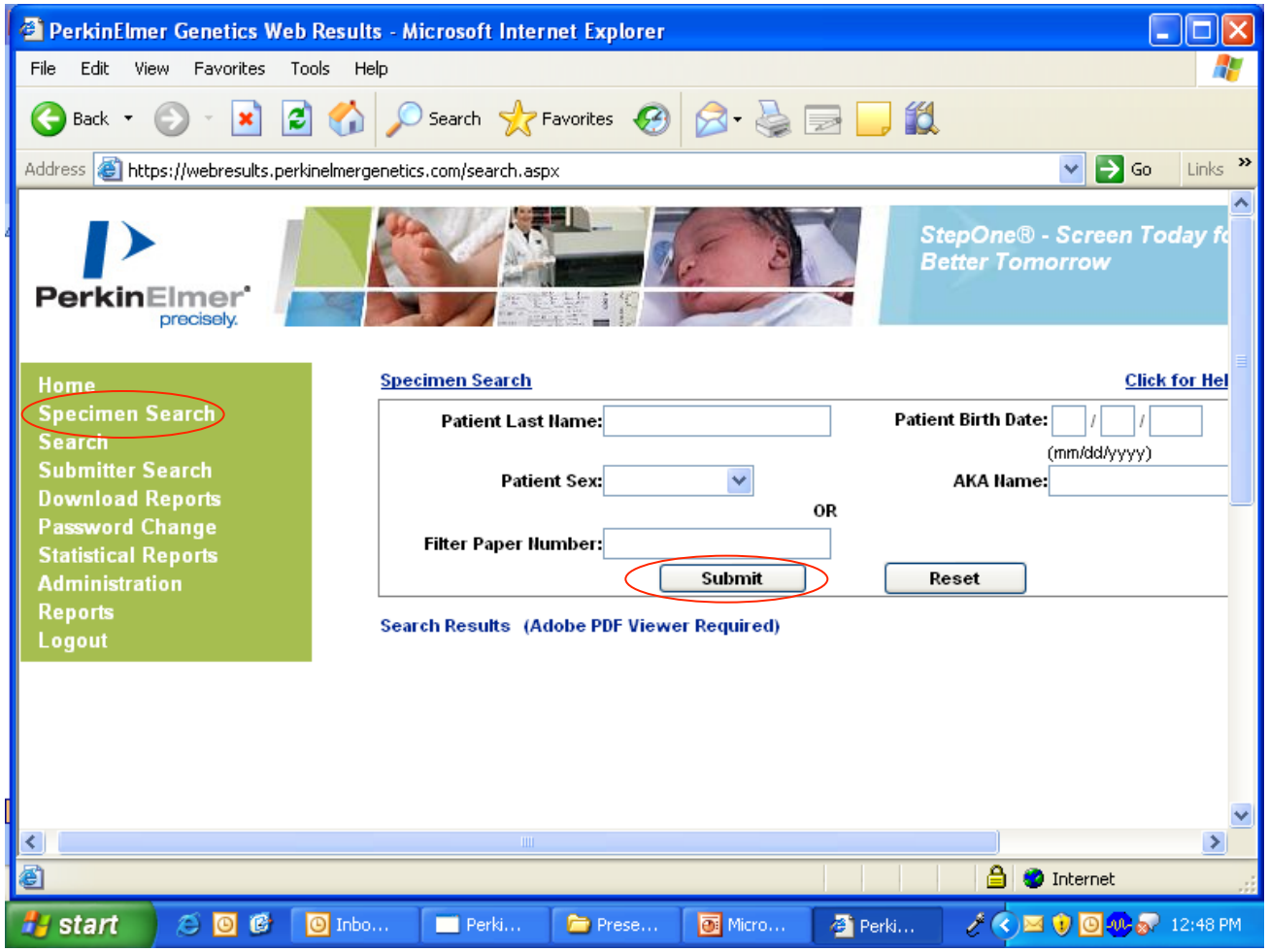

User Info - Microsoft Internet Explorer
File Edit View Favorites Tools Help
Back Search Favorites
Address http://2s3kpssweb02/userinfo.aspx
Go Links
PerkinElmer precisely.
StepOne® - Screen Today for a Better Tomorrow
Test Mode
Home
User Info
Specimen Search
Search
Submitter Search
Download Reports
Password Change
Statistical Reports
Administration
Reports
Logout
User Info
Click for Help
UserID: sfelinczak
Email: SUSAN.FELINCZAK@PE
Last Name: Felinczak
First Name: Susan
Practice/Sub Name: PerkinElmer Genetics
Address One: 90 Emerson Lane
City: Bridgeville
Address Two:
State: Pennsylvania
Address Three:
Zip: 15017
Address Four:
Country: United States
Phone Number: 412-220-2300
Fax Number:
Update Info
Security Questions Update
Question One:
What is your mother's maiden name?
Answer:
Question Two:
Answer:
Question Three:
Answer:
tally
What is your mother's maiden name?
What street did you grow up on?
What was the name of your first pet?
What was the last name of your first grade teacher
What are the last four digits of your social security
What was your first telephone number?
Update Questions
Local intranet
start
Microsoft Office Com...
User Info - Microsoft ...
2:15 PM

Results Portal – Home Page



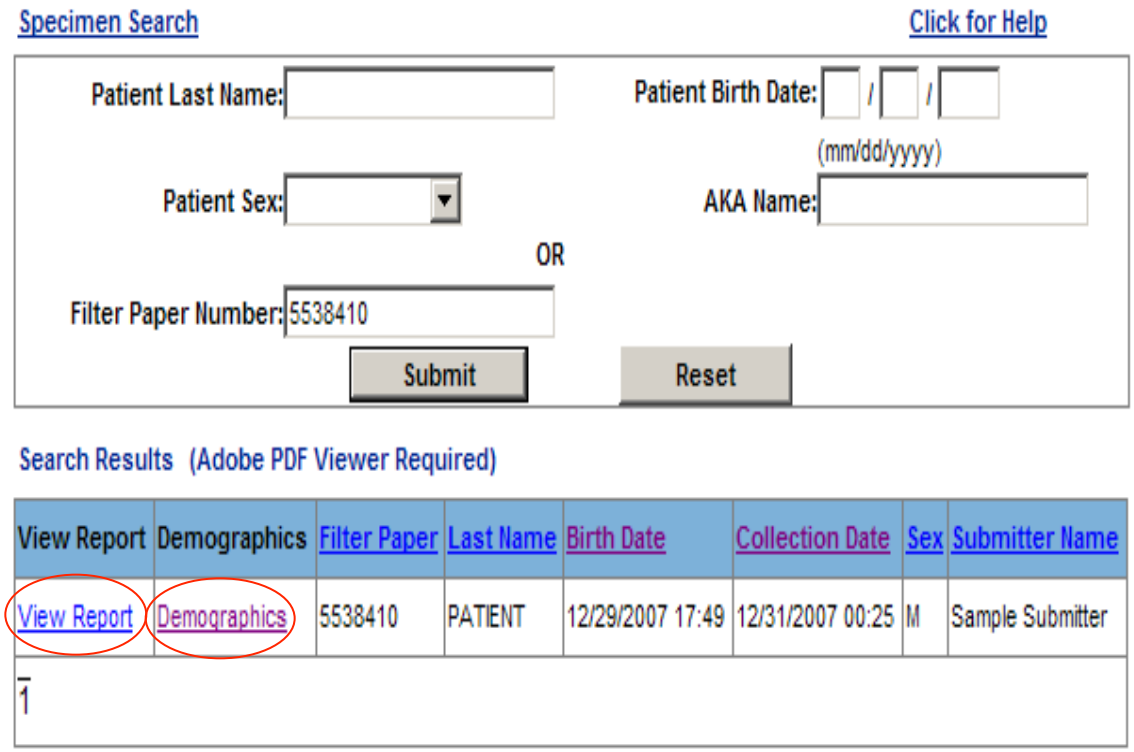
- Your **"Home"** page will be based upon your user rights which is either Search, Download or both.
- For example, if your access is for **"Search"** only the **"Specimen Search"** screen will launch directly to your **"Home"** page.
- The following functions are also accessible by clicking the links located on the left navigation menu which is user based:
  - > **Downloading Reports**
  - > **Password Change**
  - > **Statistical Reports**
  - > **Reports**
  - > **Logout**

Results Portal – Search Module



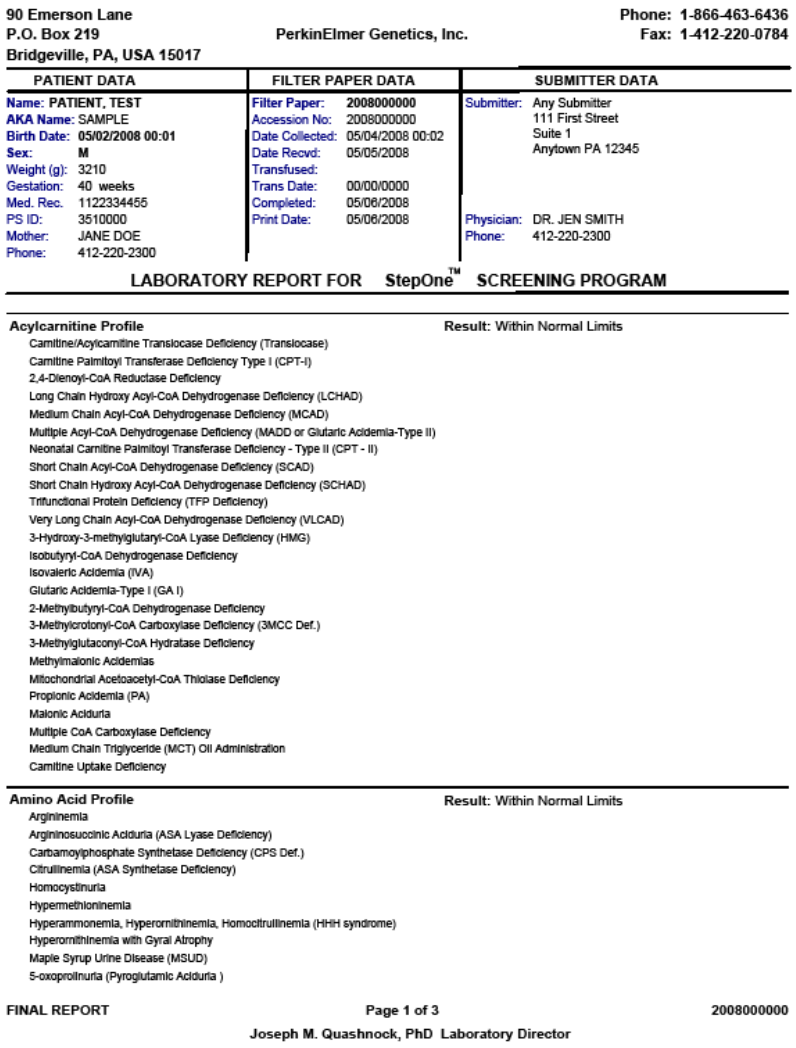
- The **"Specimen Search"** screen will show the criteria that you can use to search for newborns in our database. You can enter information into one or all fields.
- Once you enter the criteria, click the **"Submit"** button.

Results Portal – Search Module



- The box shown to the left has the results for your search.
- This screen will allow you to review the **"Report"** or the **"Demographic"** information for the newborn.
- To view the test results, click on the **"View Report"** link.
- To view the newborn's information, click on the **"Demographics"** link.

Results Portal – Search Module



90 Emerson Lane
P.O. Box 219
Bridgeville, PA, USA 15017

PerkinElmer Genetics, Inc.

Phone: 1-866-463-6436
Fax: 1-412-220-0784

PATIENT DATA		FILTER PAPER DATA		SUBMITTER DATA	
Name:	PATIENT, TEST	Filter Paper:	2008000000	Submitter:	Any Submitter
AKA Name:	SAMPLE	Accession No:	2008000000		111 First Street
Birth Date:	05/02/2008 00:01	Date Collected:	05/04/2008 00:02		Suite 1
Sex:	M	Date Recvd:	05/05/2008		Anytown PA 12345
Weight (g):	3210	Transfused:			
Gestation:	40 weeks	Trans Date:	00/00/0000		
Med. Rec.	1122334455	Completed:	05/06/2008		
PS ID:	3510000	Print Date:	05/06/2008	Physician:	DR. JEN SMITH
Mother:	JANE DOE			Phone:	412-220-2300
Phone:	412-220-2300				

LABORATORY REPORT FOR StepOne™ SCREENING PROGRAM

Acylcarnitine Profile — Result: Within Normal Limits

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
Carnitine Palmitoyl Transferase Deficiency Type I (CPT-I)
2,4-Dienoyl-CoA Reductase Deficiency
Long Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHAD)
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidemia-Type II)
Neonatal Carnitine Palmitoyl Transferase Deficiency - Type II (CPT - II)
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
Trifunctional Protein Deficiency (TFP Deficiency)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
3-Hydroxy-3-methylglutaryl-CoA Lyase Deficiency (HMG)
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia (IVA)
Glutaric Acidemia-Type I (GA I)
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Def.)
3-Methylglutaconyl-CoA Hydratase Deficiency
Methylmalonic Acidemias
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
Propionic Acidemia (PA)
Malonic Aciduria
Multiple CoA Carboxylase Deficiency
Medium Chain Triglyceride (MCT) Oil Administration
Carnitine Uptake Deficiency

Amino Acid Profile — Result: Within Normal Limits

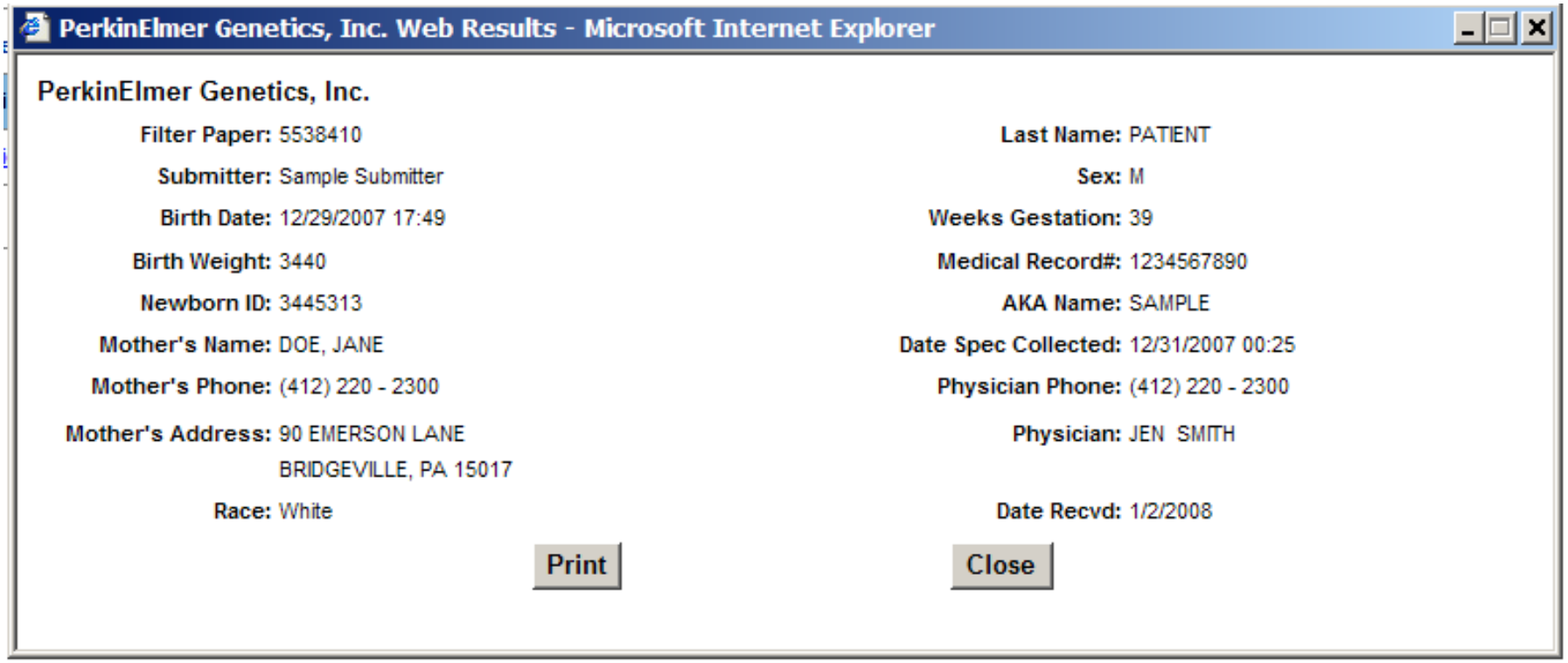
Argininemia
Argininosuccinic Aciduria (ASA Lyase Deficiency)
Carbamoylphosphate Synthetase Deficiency (CPS Def.)
Citrullinemia (ASA Synthetase Deficiency)
Homocystinuria
Hypermethioninemia
Hyperammonemia, Hyperornithinemia, Homocitrullinemia (HHH syndrome)
Hyperornithinemia with Gyral Atrophy
Maple Syrup Urine Disease (MSUD)
5-oxoprolinuria (Pyroglutamic Aciduria)

FINAL REPORT — Page 1 of 3 — 2008000000

Joseph M. Quashnock, PhD Laboratory Director

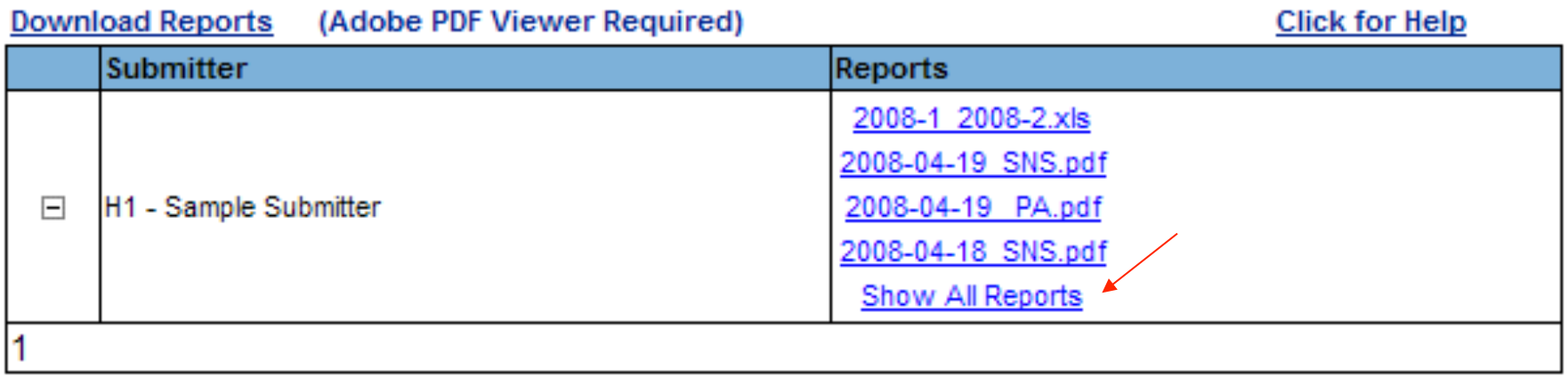
- By selecting **"View Report"** a newborn report will be generated like the view shown to the left.
- This report can be printed from the browser by selecting **FILE → Print.**

Results Portal – Search Module



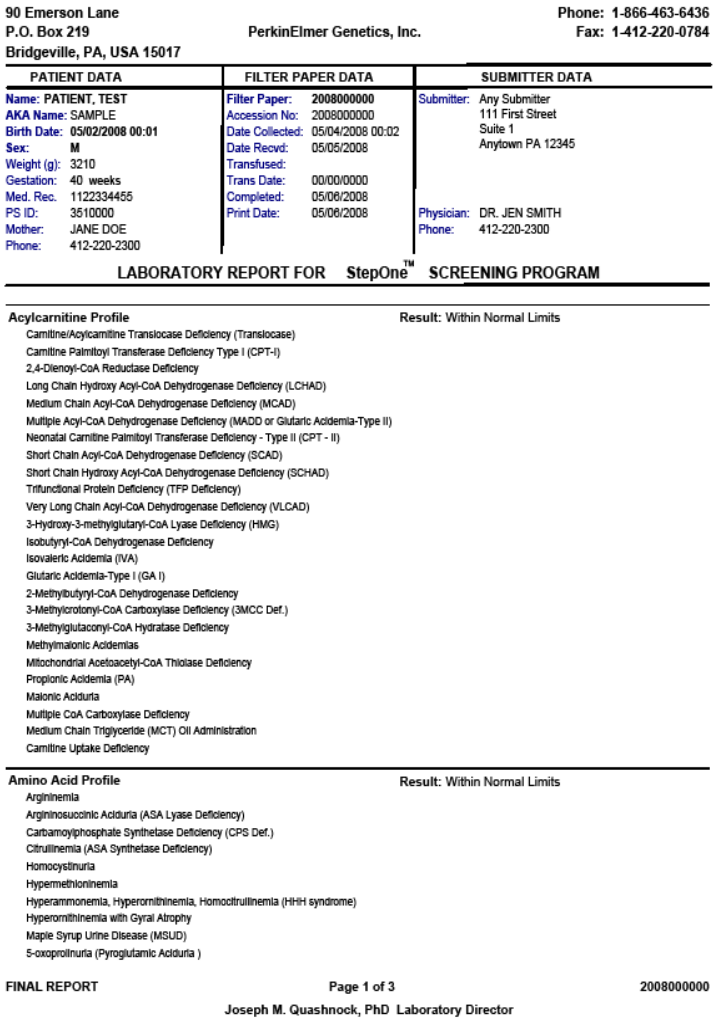
- By selecting, **"Demographics"** the system will launch the following screen which will provide you with all information supplied to PerkinElmer Genetics regarding the specified newborn.

Results Portal – Download Module



- To download a series of reports and view them through Adobe, click on the **"Download Report"** option on the menu bar located to the left of your screen.
- The screen will list the most recent reports and provide a link to **"Show All Reports"**.
- In the download file layout you will see a list of files by date that are available for download.
- The files are dated as to when the reports are released.
- Reports are posted each day by 4 a.m.
- Click the date link to view the reports, this will launch an Adobe viewer. Adobe Acrobat Viewer is required from www.adobe.com

Results Portal – Download Module



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Phone: 1-866-463-6436
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PATIENT DATA		FILTER PAPER DATA		SUBMITTER DATA	
Name:	PATIENT, TEST	Filter Paper:	2008000000	Submitter:	Any Submitter
AKA Name:	SAMPLE	Accession No:	2008000000		111 First Street
Birth Date:	05/02/2008 00:01	Date Collected:	05/04/2008 00:02		Suite 1
Sex:	M	Date Recvd:	05/05/2008		Anytown PA 12345
Weight (g):	3210	Transfused:			
Gestation:	40 weeks	Trans Date:	00/00/0000		
Med. Rec.	1122334455	Completed:	05/06/2008		
PS ID:	3510000	Print Date:	05/06/2008	Physician:	DR. JEN SMITH
Mother:	JANE DOE			Phone:	412-220-2300
Phone:	412-220-2300				

LABORATORY REPORT FOR StepOne™ SCREENING PROGRAM

Acylcarnitine Profile — **Result:** Within Normal Limits

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
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Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
Trifunctional Protein Deficiency (TFP Deficiency)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
3-Hydroxy-3-methylglutaryl-CoA Lyase Deficiency (HMG)
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia (IVA)
Glutaric Acidemia-Type I (GA I)
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Def.)
3-Methylglutaconyl-CoA Hydratase Deficiency
Methylmalonic Acidemias
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
Propionic Acidemia (PA)
Malonic Aciduria
Multiple CoA Carboxylase Deficiency
Medium Chain Triglyceride (MCT) Oil Administration
Carnitine Uptake Deficiency

Amino Acid Profile — **Result:** Within Normal Limits

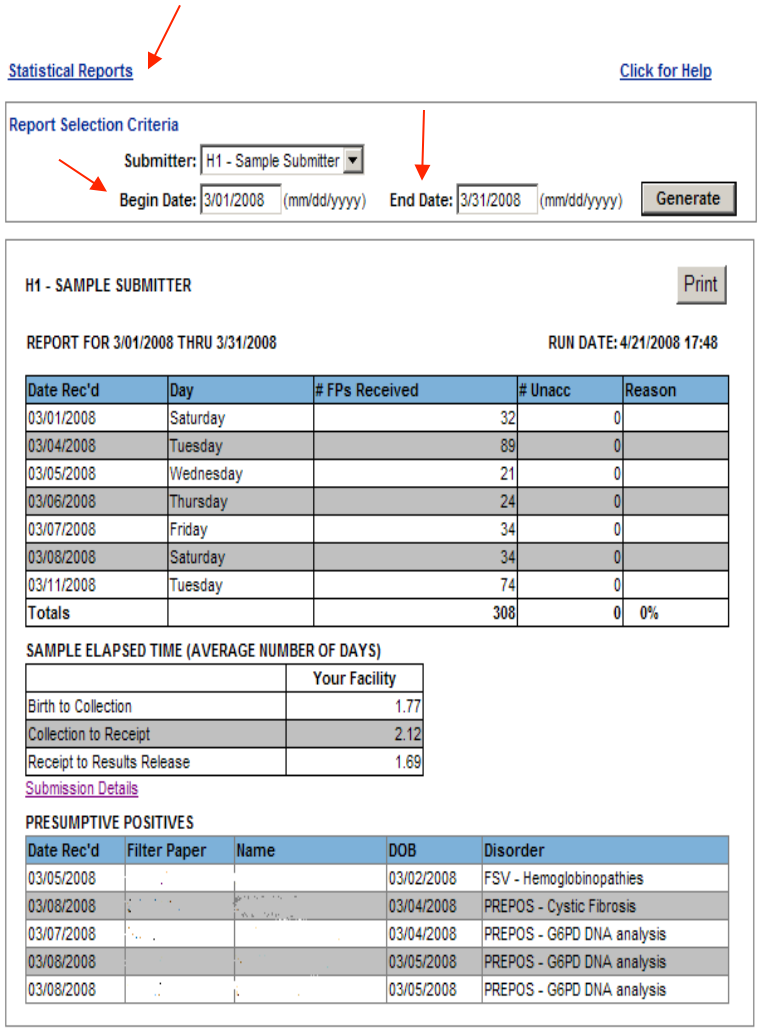
Argininemia
Argininosuccinic Aciduria (ASA Lyase Deficiency)
Carbamoylphosphate Synthetase Deficiency (CPS Def.)
Citrullinemia (ASA Synthetase Deficiency)
Homocystinuria
Hypermethioninemia
Hyperammonemia, Hyperornithinemia, Homocitrullinemia (HHH syndrome)
Hyperornithinemia with Gyral Atrophy
Maple Syrup Urine Disease (MSUD)
5-oxoprolinuria (Pyroglutamic Aciduria)

FINAL REPORT Page 1 of 3 2008000000

Joseph M. Quashnock, PhD Laboratory Director

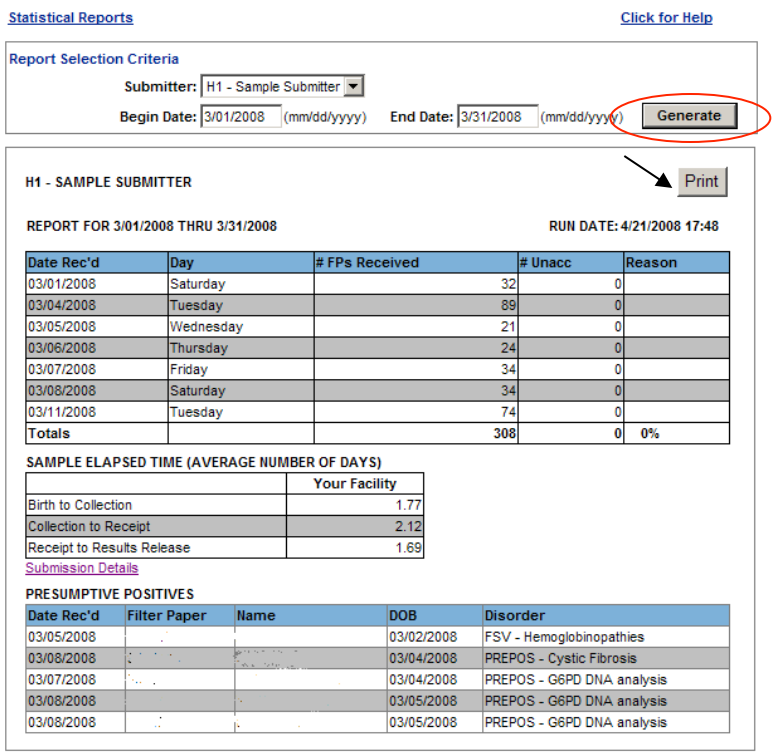
- A series of reports will generate like the view shown to the left. The number of reports in the series will depend upon how many were released that day.
- If you select **"Print"**, all reports in the file will print.

Results Portal – Statistical Reports Module



- By clicking on the **"Statistical Reports"** option located on the left navigation menu, the screen at the left will launch.
- Select a date range by clicking in the **"Begin Date"** and **"End Date"** fields. Dates can be changed manually or by selecting them from the popup calendar.
- Submitter Specific Metrics
- Turn-around time details extracted to Excel.

Results Portal – Statistical Reports Module



Statistical Reports Click for Help

Report Selection Criteria

Submitter: H1 - Sample Submitter

Begin Date: 3/01/2008 (mm/dd/yyyy) End Date: 3/31/2008 (mm/dd/yyyy) Generate

Print

H1 - SAMPLE SUBMITTER

REPORT FOR 3/01/2008 THRU 3/31/2008 RUN DATE: 4/21/2008 17:48

Date Rec'd	Day	# FPs Received	# Unacc	Reason
03/01/2008	Saturday	32	0	
03/04/2008	Tuesday	89	0	
03/05/2008	Wednesday	21	0	
03/06/2008	Thursday	24	0	
03/07/2008	Friday	34	0	
03/08/2008	Saturday	34	0	
03/11/2008	Tuesday	74	0	
Totals		308	0	0%

SAMPLE ELAPSED TIME (AVERAGE NUMBER OF DAYS)

	Your Facility
Birth to Collection	1.77
Collection to Receipt	2.12
Receipt to Results Release	1.69

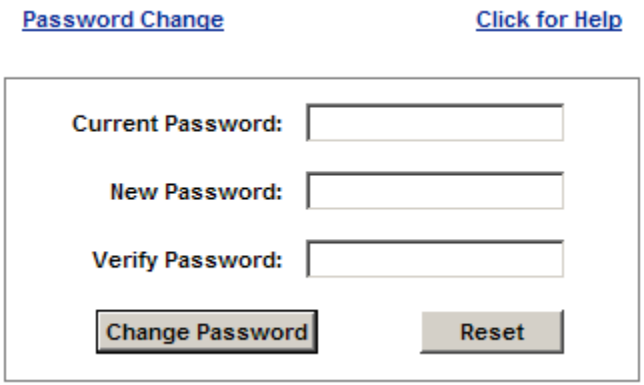
Submission Details

PRESUMPTIVE POSITIVES

Date Rec'd	Filter Paper	Name	DOB	Disorder
03/05/2008			03/02/2008	FSV - Hemoglobinopathies
03/08/2008			03/04/2008	PREPOS - Cystic Fibrosis
03/07/2008			03/04/2008	PREPOS - G6PD DNA analysis
03/08/2008			03/05/2008	PREPOS - G6PD DNA analysis
03/08/2008			03/05/2008	PREPOS - G6PD DNA analysis

- Once you have defined the date criteria, click on **"Generate"** and the results of your selection will launch providing you with the following information:
  - Summary and detail of all specimens submitted
  - Number of unacceptable samples
  - Turn-Around times
  - Presumptive positives
- This report can be printed by clicking on the **"Print"** button.

Results Portal – Password Change



- The system will prompt you to change your password every 90 days for increased security.
- Cannot re-use previous password
- Letter and number combination
- Password resets or locked accounts please contact Client Services at (866) 463-6436