California Prenatal Screening Program

Kaiser Permanente
Genetic Screening Services
Presented by:
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Genetic Counselor, Genetics Department
Baldwin Park Medical Center

Time to Change

- The Expanded AFP Screening Program will become the California Prenatal Screening Program (CPSP)
  - Will expand to include screening in the first trimester
  - Now an integrated screening approach
  - Starting on March 12, 2009
What Will Change?

- Currently, 1st trimester specimens are sent to Quest, and 2nd trimester specimens are sent to the State-funded Newborn and Prenatal Screening (NAPS) lab at Kaiser.
- Beginning 3/12/09, ALL maternal serum screening specimens will be analyzed at the Kaiser NAPS lab.
- New 1st and 2nd trimester lab forms from the State

Prenatal Screening: State Regulations

- All pregnant women must be offered participation in California’s maternal serum screening program. Patients have the right to decline screening.
- As of 4/1/09, no lab, business, or program can perform prenatal screening for chromosome abnormalities and neural tube defects other than the CA Prenatal Screening Program administered by the CA Dept. of Public Health.
- Kaiser pays the State $162 per patient. It is a one-time fee which covers either one or two blood specimen results plus genetic counseling and diagnostic services (ultrasound, CVS, amniocentesis) if the result is Screen Positive.
### Which Conditions Does CPSP Screen For?

- Continues to screen for the same four genetic conditions/birth defects:
  - Down syndrome
  - Trisomy 18
  - Neural tube defects/abdominal wall defects
  - Smith-Lemli-Opitz syndrome

- If patient considering diagnostic testing (CVS/amnio), refer to Genetics

### CPSP Offers 3 Types of Screening Tests

- **Quad marker screening**
  - One blood draw at 15-20 weeks

- **Serum integrated screening**
  - Combines 1<sup>st</sup> trimester blood test results with 2<sup>nd</sup> trimester blood test results

- **Full integrated screening**
  - Combines nuchal translucency (NT) results with 1<sup>st</sup> and 2<sup>nd</sup> trimester blood test results
  - Preliminary risk assessment available in 1<sup>st</sup> trimester
CA Prenatal Screening Program

1st trimester specimen drawn
10w0d – 13w6d

*Down syndrome detection rates

No NT

NT obtained
11w2d – 14w2d

2nd T specimen NOT drawn

2nd T specimen drawn
15w0d – 20w0d

NO Risk Assessment

Serum Integrated Screening (85%)*

1st T Risk Assessment (75%)*

Full Integrated Screening (90%)*

Quad Screening (80%)*

Quad + NT Screening (89%)*

2nd T specimen drawn
(no 1st T specimen)

1st T Risk Assessment

Timing

- 1st Trimester Blood Draw:
  10w0d – 13w6d

- Nuchal Translucency (NT):
  11w2d – 14w2d (CRL 45mm-84mm)

- 2nd Trimester Blood Draw:
  15w0d – 20w0d
### Program Analytes

<table>
<thead>
<tr>
<th>1(^{st}) trimester:</th>
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<tbody>
<tr>
<td>• PAPP-A (pregnancy associated plasma protein A)</td>
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<tr>
<td>• hCG (human chorionic gonadotropin)</td>
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<tr>
<td>• Nuchal Translucency (NT) if available</td>
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</table>

<table>
<thead>
<tr>
<th>2(^{nd}) trimester:</th>
</tr>
</thead>
<tbody>
<tr>
<td>• AFP (alpha-fetoprotein)</td>
</tr>
<tr>
<td>• hCG (human chorionic gonadotropin)</td>
</tr>
<tr>
<td>• uE3 (unconjugated estriol)</td>
</tr>
<tr>
<td>• INH (inhibin)</td>
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### Nuchal Translucency (NT)

<table>
<thead>
<tr>
<th><img src="image.jpg" alt="Image" /></th>
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<tbody>
<tr>
<td>• Ultrasound measurement of fluid-filled space at the back of the fetal neck</td>
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<tr>
<td>• Increased NT is associated with increased risk for chromosome abnormalities, heart defects, and other birth defects</td>
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<tr>
<td>• Must be done by certified NT practitioner approved by the CA Prenatal Screening Program</td>
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<td>• Cost is not reimbursed by the State</td>
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Nuchal Translucency (NT)

1 mm

3 mm

Quad Marker Screening

- Blood draw at 15w0d-20w0d.
- Should be offered to:
  - Women who did not have a 1st trimester blood specimen sent to the State
  - Women who had CVS → risk assessment for NTD/SLOS only
- Quad + NT Screening:
  - Can provide a 1st trimester NT to obtain a higher detection rate for Down syndrome and Trisomy 18.
- Results:
  - Screen Negative
  - Screen Positive → Patient referred to Genetics by PNS Coordinator. State will cover cost of genetic counseling, ultrasound, and amnio.
### Serum Integrated Screening (No NT)

**1ST TRIMESTER:**
- Blood draw: 10w0d-13w6d
- State sends OB provider and patient an acknowledgement letter after receiving 1st trimester blood specimen. No preliminary results provided.

**2ND TRIMESTER:**
- Blood draw: 15w0d-20w0d
- Results provided after 2nd trimester blood draw.
  - Screen Negative
  - Screen Positive → Patient referred to Genetics by PNS Coordinator. State will cover cost of genetic counseling, ultrasound, and amnio.

### Full Integrated Screening

**FIRST TRIMESTER:**
- Blood draw: 10w0d-13w6d
- NT: 11w2d-14w2d

**First Trimester Screening Results:**
- “Preliminary Risk Assessment” (Screen Negative) → Draw 2nd trimester specimen for full integrated result
- Screen Positive → Patient referred to Genetics by PNS Coordinator
- Preliminary result identifies patients at highest risk
Options for Patients with Positive FTS Results

Refer patient to Genetics for follow-up

Genetic Counseling

Patient declines follow-up services

CVS (1st trimester)

Amnio and US (15-20 w)

Patient elects combined risk assessment

Patient elects combined risk assessment

Patient declines combined risk assessment

Obtain 2nd trimester specimen

Obtain 2nd trimester specimen

Obtain 2nd trimester specimen

Full Integrated Screening (cont.)

SECOND TRIMESTER:

- Blood draw: 15w0d-20w0d
- Full Integrated Screening Results:
  - Screen Negative
  - Screen Positive → Patient referred to Genetics by PNS Coordinator. State will cover cost of genetic counseling, ultrasound, and amnio.
Special Circumstances: Multiple Gestation

- Twin pregnancies can have quad, serum integrated, or full integrated screening.

<table>
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<tr>
<th>Type</th>
<th>1st Trimester</th>
<th>2nd Trimester</th>
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<tr>
<td>Quad</td>
<td>T21, NTD</td>
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<tr>
<td>Quad + NT</td>
<td>T21, T18, NTD</td>
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<tr>
<td>Serum Integrated</td>
<td>T21, NTD</td>
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<tr>
<td>Full Integrated</td>
<td>T21, T18, NTD</td>
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<tr>
<td>1st Trimester: Preliminary Risk</td>
<td>T21, T18</td>
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- Serum screening is not available for pregnancies with 3 or more fetuses. Offer NT only.

Special Circumstances: Fetal Demise or Reduction

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<tr>
<th>Scenario</th>
<th>1st Trimester Specimen</th>
<th>2nd Trimester Specimen</th>
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<tr>
<td>Fetal Demise ≤ 8w0d</td>
<td>Do NOT draw specimen</td>
<td>Screenable* (quad only)</td>
</tr>
<tr>
<td>Fetal Demise &gt; 8w0d</td>
<td>NOT Screenable</td>
<td>NOT Screenable</td>
</tr>
<tr>
<td>Fetal Reduction</td>
<td>NOT Screenable</td>
<td>NOT Screenable</td>
</tr>
<tr>
<td>(Anytime During Pregnancy)</td>
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*Write down number of VIABLE fetuses on lab form.
Integrated Screening in Kaiser Permanente

What is the process?

1st Trimester Blood Draw

- Order 1st blood draw in Health Connect: 1ST TRIMESTER SCREEN, CALIFORNIA (PAPP-A, HCG) insert order code here
- If 2nd trimester draw also being ordered, place as future order (expected = 16 weeks GA)
- Complete State 1st trimester lab form – BLUE border
- Peel off label from 1st trimester lab form and place on 2nd trimester lab form
- Document 1st trimester form number in HealthConnect
- Optional: Contact first trimester screening (FTS) case manager to arrange NT appointment
2nd Trimester Blood Draw

- Order 2nd blood draw in HealthConnect: 2ND TRIMESTER SCREEN, CALIFORNIA (AFP, HCG, UE3, INHIBIN) Insert order code here
- Complete State 2nd trimester lab form – GREEN border
- Make sure label from 1st trimester lab form is placed on 2nd trimester lab form
  - If label is not available, write in the 1st trimester form number

CA Prenatal Screening Consent Form
1st Trimester Lab Form

2nd Trimester Lab Form
Nuchal Translucency (NT) Information

- Complete this section if the patient already had NT
- NT practitioner credential number is required
- Provide NT and CRL measurements in mm
- For twin pregnancies:
  - Complete CRL and NT measurements for each twin
  - Indicate chorionicity

Pregnancy Dating

- Complete only ONE dating method
- US dating is preferred
  - Use BPD or CRL (do not use average, FL, or AC)
  - Use decimal OR weeks + days (not both)
- Only use LMP if US dating is not available
- Never use physical exam
- Do not use LMP for twin pregnancies
CVS Question on 2nd Trimester Form

- Mark “yes” ONLY if the patient had CVS with fetal karyotype
- If marked “yes,” patient will receive NTD/SCD only screening

Question Regarding Research

- If the patient declines the use of her specimen for research on the consent form, fill in the appropriate box on the lab forms.
  - First Trimester Form → Question 13
  - Second Trimester Form → Question 15

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13. HAS PATIENT HAD CVS?

- YES
- NO

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15. PATIENT CONSENT FORM:

If patient marked "decline the use of my specimen for research" on the consent form, fill in the box at right:
- YES
- NO
First Trimester Specimen Label

1. Peel QR T
2. Apply to Part A of 1st trimester form

Label is on PINK page of 1st trimester form

Second Trimester Screening

15 WEEKS 0 DAYS TO 20 WEEKS 6 DAYS
CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
CALIFORNIA PRENATAL SCREENING PROGRAM
(866) 718-9715 Toll Free

PLEASE WRITE CLEARLY USE CAPITAL LETTERS.

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Did you know?

- 30% of all Prenatal Screening forms are submitted with missing or inaccurate patient information.

Here is what you and your staff can do to improve the quality and accuracy of your patients' Prenatal Screening results:

- Complete every question on the test request form (TRF).
  - If you're unsure whether to request a procedure, ask a question.
  - Ask a question even if you're unsure.

- Don't forget to include an accurate BIRTHDATE in the record. If the patient's age is unknown, you've got a chance to get it right.

- Complete the TRF current, accurate, and as complete as possible.

- Only submit a TRF if the patient is pregnant or has a risk factor.

- Always check the patient's address and all other relevant information.

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Genetic Screening Services

- Prenatal Screening (PNS) Coordinators:
  Caleen A. Danon, MS
  Aki Logg, MS, CGC
  Amy Liou, MS, CGC

- Prenatal Screening Support Staff:
  Rachiel Reyes
  John Criscione

Phone: (626) 564-3322
Tie Line: 8-338-3322

Prenatal Screening Case Manager Contact List

<table>
<thead>
<tr>
<th>Medical Center</th>
<th>Case Manager</th>
<th>Phone number</th>
<th>Tie line</th>
</tr>
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<tbody>
<tr>
<td>Baldwin Park</td>
<td>Stacy Perez</td>
<td>626-851-6587</td>
<td>370</td>
</tr>
<tr>
<td>Bellflower</td>
<td>Shirley Rone</td>
<td>562-657-4055</td>
<td>327</td>
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<tr>
<td>South Bay</td>
<td>Shirley Rone</td>
<td>562-657-4055</td>
<td>327</td>
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<tr>
<td>Los Angeles</td>
<td>Traci Richardson</td>
<td>323-783-8669</td>
<td>363</td>
</tr>
<tr>
<td>West LA</td>
<td>Traci Richardson</td>
<td>323-783-8669</td>
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<tr>
<td>Riverside</td>
<td>Cindy Williams</td>
<td>951-353-5359</td>
<td>258</td>
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<tr>
<td>Fontana</td>
<td>Tyra Blount</td>
<td>909-427-3109</td>
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<tr>
<td>Anaheim/OC</td>
<td>Vickylynn Rubio</td>
<td>949-932-5654</td>
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<tr>
<td>San Diego</td>
<td>Laura Diaz</td>
<td>619-528-7113</td>
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<tr>
<td>Panorama City</td>
<td>Araceli Rodriguez</td>
<td>818-375-2776</td>
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<tr>
<td>Woodland Hills</td>
<td>Araceli Rodriguez</td>
<td>818-375-2776</td>
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<tr>
<td>Kern County</td>
<td>Sonia Dhillon</td>
<td>661-852-3606</td>
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