Western states public health genetics and newborn screening leaders’ perceptions of the Western States Genetic Services Collaborative: Report of key informant interview findings

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Purpose
As one of seven Genetic Service and Newborn Screening Regional Collaboratives established with grant funds from the Genetic Services Branch, Maternal Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA), the Western States Genetic Services Collaborative (WSGSC) works to strengthen and support the genetics and newborn screening (NBS) capacity of Alaska, California, Hawaii, Idaho, Oregon, Washington and Guam. The purpose of the Genetic Services Collaboratives is to use a regional approach to address state gaps in genetic services and resources. A fundamental goal of the national effort is to improve the health of children and families by bringing services closer to local committees and promoting translation of genetic medicine into public health and healthcare services.1

A primary goal of the WSGSC is to increase the capacity of collaborating states’ and territory’s public health agencies to perform genetic services assessment, policy development, and assurance functions. State/territory leaders in public health genetics and newborn screening are regular participants in Collaborative planning processes, annual meetings, Steering Committee, work groups and other Collaborative projects. As part of a larger Evaluation Plan, the WSGSC seeks to assess to what extent and in what ways public health capacity is enhanced by Collaborative activities. Understanding perceptions of state public health genetics leaders related to these questions is a crucial means of evaluating how the goal of capacity building is being met by Collaborative activities. Key informant interviews of state/territory public health genetics and newborn screening leaders are conducted each year as part of the Western States Genetic Services Regional Collaborative Evaluation Plan. Key informant interviews were conducted in 2007, 2008, and in May and June of 2009. This report includes findings from the 2009 interviews.

Methods
Individuals who attend an annual WSGSC “Regional Summit” include family representatives or advocates, genetic counselors, medical geneticists, public health professionals from state genetic services and newborn screening programs, and other health care providers. These individuals, whose attendance at the Regional Summit may vary from year to year, are considered to be stakeholders in the WSGSC. The WSGSC
The evaluation team elected to concentrate on interviewing only public health stakeholders due to budget limitations and the public health focus of the grants. Twelve genetics services and newborn screening public health professionals (including the WSGSC Co-Principal Investigators) from the six western states (AK, CA, HI, ID, OR, WA) and Guam were asked to participate in open-ended, semi-structured telephone interviews. Eight individuals representing state public health agencies in AK, HI, ID, OR and WA agreed to participate. The WSGSC evaluator conducted the interviews. Interview times ranged from forty to sixty minutes. One interviewee submitted written responses to the open-ended questionnaire. Prior to scheduling the interview, interviewees received a copy of the questionnaire and a copy of the guiding philosophies of the WSGSC.

The interview instrument, based upon a theoretical model that describes collaborative relationships\(^2\) was adapted in 2008 and again in 2009 from an instrument developed in 2006 by the WSGSC Evaluation Advisory Committee. Interview questions were written to measure outcomes of interest from the WSGSC Logic Model, including interviewee perceptions of what their states both “get” and “give” by participating in the Collaborative. The concept of “get” and “give” has been used in planning and negotiating grant activities among the states.

Table 1 depicts aspects of the theoretical model that was used to structure the interview instrument. The evaluation team sought to assess interviewee perceptions of WSGSC functioning in terms of processes that characterize collaborative relationships. The instrument also included a question to assess whether key informants believed the WSGSC adheres to Guiding Principles earlier set forth by the entire Collaborative. The interviewer told interviewees their comments would be aggregated with those of other interviewees and included in a report to the Maternal Child Health Bureau.

### Table 1. Theoretical model used as a basis for Western States Genetic Services Collaborative public health professional key informant interviews that assessed respondent beliefs about Collaborative functioning.

<table>
<thead>
<tr>
<th>Integration</th>
<th>Process</th>
<th>Structure</th>
<th>Purpose</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Contribution</td>
<td>Support group</td>
<td>Mutual exchanges to support each other’s efforts. Build mutual obligation and trust.</td>
</tr>
<tr>
<td></td>
<td>Coordination</td>
<td>Task force, council, alliance</td>
<td>Match and coordinate needs, resources, and activities. Limit duplication of services. Adjust current activities for more efficient and effective results.</td>
</tr>
<tr>
<td></td>
<td>Cooperation</td>
<td>Partnership, consortium, coalition</td>
<td>Link resources to help parties achieve joint goals. Discover shared interests. Build trust by working together.</td>
</tr>
<tr>
<td>High</td>
<td>Collaboration</td>
<td>Collaborative</td>
<td>Develop shared vision. Build inter-dependent system to address issues and opportunities. Share resources.</td>
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Results

Get

1. What new information about genetic services (including NBS) have you learned from participating in the Collaborative?

Genetics
- General news and updates on what other public health genetics and newborn screening programs are doing. (3)
- Gained information about how telemedicine is working for other states. (2)
- Learn what other states are doing with regard to measuring outcomes and performance measures.
- Usefulness of information learned varies from state to state, what is useful for one public health program might not be useful for another.
- Did not get much new information from 2008 Regional Summit.
- Nothing new.
- Funding opportunities.
- National policy initiatives.
- Time devoted to discussing reimbursement issues and licensing issues are not of interest to our program. Our state does not spend time talking to providers about reimbursement, including public health insurance (Medicaid).
- I would love to get a mandate in our state for insurance coverage for medically necessary food, but I’m not allowed to open this conversation because that interferes with private business.
- I am struggling to find value in the WSGSC.

Newborn Screening
- Appreciated discussion of uncommon disorders. (3)
- At the April NBS meeting learned what other states are doing with regard to NBS practices. (2)
- Appreciated discussion of cut off levels. (2)
- Would like to see more work such as April NBS meeting that contains concrete discussions and shows direct benefit. (2)
- Learned about national efforts in NBS and the long-term newborn screening (NBS) database. (2)
- Important to discuss information about VLCAD condition.
- Glad one outcome of April NBS meeting is that practitioners will pool data and a publication may result.
- Learned about and shared information about new mutations, what they are, what to do about them in terms of treatment.
- Was good to see physicians becoming excited about discussions at the April NBS meeting.
- Would prefer to just be in the Northwest Regional Newborn Screening collaboration rather than the WSGSC.

2. What assistance have you received by participating in the Collaborative?

Genetics
- None. (4)
- Funding to help with increasing access to clinical services. (3) (Note: One respondent indicated outreach would be funded through MCHB block grant if WSGSC funds not available.)
- Agreement with Hawaii to provide incentives for billing and reimbursement study.
• Funding to pay for provider and family training DVD.
• Help with evaluation of services.
• Communication with other collaborators get a fuller picture of access issues.

**Newborn Screening**
• Direct benefit from information shared at April NBS meeting.

3. **What are some examples of beneficial networking that have occurred for you or your agency from Collaborative activities?**

**Genetics**
• An opportunity to meet with genetics and newborn screening people from other states. (2)
• Networking within state by updating genetics related materials for state medical home website.
• Discussions and learning from other states helps us improve our existing programs.
• Gives us a chance to get together with other people in our own states who we typically may not be able to meet with.

**Newborn Screening**
• Difficult to identify differences between WSGSC activities in NBS and Northwest Regional NBS group. (3)
• Connections with NBS heads in other states is helpful in terms of sharing ideas, getting to know the people. (3)
• An opportunity to identify what projects other states in the region may want to pursue. (2)
• Helpful to hear what other states do to respond to specific conditions, what their outcomes of certain conditions are, and actions they take for rare conditions. (2)
• Discussing with collaborators from other states what is working and what is not working in NBS.
• Region needs to study whether our follow-up steps are consistent, with as little likelihood as possible of harm coming to child. Important question: “What’s the minimum we can do yet make sure the child develops normally as possible?”
• A benefit to discuss Secretary’s Advisory Council recommendations (or non-recommendations) for panel.
• No benefit.

4. **How have, or will, collaborative efforts help your state or program to meet your state goals and objectives?**

**Genetics**
• By partially funding our ability to outreach to families who live far from services and collect program data about our outreach efforts. (2)
• By studying ways to make genetics clinics self-supporting, not needing public health funding. (2)
• Helps us coordinate and share information from other states about broader MCHB program needs such as for children with special health care needs, we apply what we learn in genetics to children with special health care needs (e.g. learning about children who have a positive newborn hearing screening also have a genetic evaluation). (2)
• By augmenting what we are already doing (e.g. billing and reimbursement studies to clarify problems clinics face in billing for services).
• By providing partial funds for our training goals (DVD).
• By helping our state Title V program improve service access for children with special health care needs.
• By developing a broader awareness of adolescent transition issues.
• By developing an awareness of reimbursement issues.
• Composition of WSGSC stakeholders allows us to obtain information outside of genetics.
• Do not know.
Newborn Screening

- Have not. (2) Note: One state public health program has only NBS goals, no genetics goals.
- Same as for Question 1. Note: More meetings like the April NBS meeting would help us better educate our families because we would learn more about rare mutations, long term follow-up of rare conditions and share this information with our families.

5. What objectives of your agency were met by your sharing and coordinating resources with Collaborative members?

Genetics
- None (4)
- Improving access to genetics services. (2)
- Information sharing regarding practices and policies in other states, learning from other states mistakes or successes. (2)
- Hearing of available federal funding.

Newborn Screening
- Information sharing regarding practices and policies in other states, learning from other states mistakes or successes. (2)
- Getting ideas such as appropriate levels of follow-up for positive screens.
- None. (“This year was more vague. More specifics are needed.”)
- Information California has shared about their long term newborn screening follow-up practices has been useful.

Give

6. What assistance or resources have you extended to others in the Collaborative?

Genetics
- None (2)
- Agency leadership and staff time and expertise contributed to participation in meetings, work groups, presentations, sharing practices and knowledge. (2) (Washington-reimbursement and billing information; Hawaii-medical home expertise, adolescent transition expertise, newborn screening expertise to Guam, family representation, Clinician services (time and expertise in outreach and telehealth visits) (2)

Newborn Screening
- None (2)
- Our false positive study (on newborn screening) we helped design, told others how to do this.

7. Please describe any modifications you have made to your activities or your state’s activities to benefit others in the collaborative?

Genetics
- Conference calls with early hearing screening coordinator as a result of annual summit activities.
- Volunteer my time and expertise.
- None, but hope to eventually modify our services to include telehealth services.

Newborn Screening
- None (4)
- Modification of leadership and staff work schedules in order to participate in meetings, work groups, conference calls.
- None, but may change our metabolic newborn screening model next year.

**Mutual Benefit and Potential Challenges**

8. **Please describe activities in which you have collaborated with or developed partnerships with other Collaborative stakeholders.**

**Genetics**
- None (2)
- No active partnership, but nice to meet family representatives from our state and other states.
- No active partnership, but discussed nutrition for families with nutritionist from a different state.
- Used small amount of available WSGSC funding for existing outreach clinics as this was easiest administratively for our state.

**Newborn Screening**
- None (2)
- No active partnership, but helpful to compare specimen collection procedures with another state.

9. **Have you used findings about telemedicine and outreach clinics from the WSGSC Practice Model evaluation to plan, implement or evaluate clinical genetic services?**

**Genetics**
- Yes, stopped doing telehealth as a result. (2)
- Yes, used findings to plan or think about plans for improving access to clinical services. (2)
- No (2) and (1) We import both genetics and metabolic genetics services in to our state. Bringing people from out of state is expensive and not enough. We know our people are not getting served. Wait times are eight months to two years.

**Newborn Screening**
- No (2)

10. **From your perspective, has the collaborative adhered to the guiding principles?**

**Genetics**
- Yes (5)
- No (1)

**Newborn Screening**
- Yes
- No

11. **What conflicts of interests among Collaborators have you identified?**

**Genetics**
- None (3) and (1) Medical geneticists at times have their own idea of how things should go, with more than one in a room discussions could become contentious, but have not.
• The competition for federal grant funding is always there. (2)
• Many WSGSC participants are genetic counselors or other types of practitioners. The high amount of time spent on reimbursement issues might be self-serving.

Newborn Screening
• None (2)

12. Has the collaborative done anything that has hindered or has potential for hindering your state efforts?

Genetics
• No (5)
• One time funding could not be used to reproduce old brochures.

Newborn Screening
• No (2)

Outcomes and Quality Improvement

13. The WSGSC Outcomes Work Group developed a flexible definition of genetic services to use in measuring outcomes. It’s posted on the Collaborative web site. Have you heard about or read about this Defining Genetic Services Framework?

Genetics
• No (2)
• Yes (3)

Newborn Screening
• Not sure
• Yes

14. Has your program used the Defining Genetic Services Framework? How?

Genetics
• No (4)
• Yes – used it in planning state genetic services outcomes in terms of different levels of outcomes

Newborn Screening
• No (2) and (1) It’s not useful to me.

15. Have you heard about or read about the Genetic Services Outcomes Menu and other outcomes or quality improvement projects? Which ones?

16. Has you program used the Genetic Services Outcomes Menu? How?

Genetics
• Yes to Menu, used Menu to think through outcomes for our program Logic Model. (2)
• Yes to Menu, but do not remember thinking it is relevant to my job.
• Yes to Menu, but have not used.
• Have not heard of any.
• Yes to Mountain States Quality Improvement Project—will tell our state clinical programs about
  the Mountain States Quality Improvement measure when it becomes available.
• Yes to Mountain States Quality Improvement and would like to collaborate with other states to
  measure patient satisfaction with services, possibly using this tool.

Newborn Screening
• Yes have heard but not used as Menu is not useful because in NBS we’re at the graduate level
  compared to clinical services in genetics when it comes to measuring outcomes. We have had
  our own quality assurance system for years.
• Yes have heard but have not used. Not sure how we would use it. No one asks us to report
  this so we would not study it. We have so much to do we could not have time to collect or study
  this type of outcome. The outcomes are not quantifiable—it is too general now. Now, if I want
detailed results of agency surveys that were done, I have to specifically ask for the full results
and I still may not get them. These surveys are examples of indicators that could be used in the
Menu.

17. What outcomes does your program use to evaluate specific activities or the
program as a whole?

Genetics
• Utilization data from regional genetics clinics.
• Patient satisfaction with clinical services, wait time for appointments, MCHB performance
  measures, insurance of patients, numbers seen, where patients seen,
• We are in the process of hiring an outside evaluator to monitor our CDC genomics grant.
• One time (2007) used a measure of people living with 80 miles of a board certified genetics
  provider to look at distribution, using again for cancer services in genomics, increases in
knowledge and understanding when we’ve given presentations to public health workforce
education (education sessions on adult onset chronic like obesity, diabetes, cancer), use of our
state Medicaid paying for genetic testing services (focus on colorectal and breast ovarian
cancer, and other genetic testing).

Newborn Screening
• Washington-% that had screening, kids missed, kids diagnosed by three months of age, early
  intervention by eight months of age, number lost to follow, lab-day to day test performance,
  monitoring turn around time, workload, number of children born matches number screened and
  number of specimens, time from positive identification to diagnosis and treatment, age of child
  when they begin treatment for different conditions, depends upon diagnosis, we have system
  similar to PEAS.
• We could work toward this, the only outcome we use the most are the Title V performance
  measures, screened positive kids got treatment in timely basis, if I have to report on it, then
  we collect it.
• Number of kids that get it, how many kids get two times, transit times of blood spot from
  hospital to lab, number of unsatisfactory specimens, demographic data, time from diagnosis to
  treatment

18. What health outcomes does your program use to evaluate specific activities or
the program as a whole?

Genetics
• None (5)
Newborn Screening

- Kids that have died, varies by diagnosis. For PKU, sickle cell and other hemoglobinopathies, congenital hypothyroid, we fund clinics to send us reports that contain various outcomes such as blood levels, number of hospitalizations, medicine compliance, neurological test results, cognitive tests, motor development-we and the clinics monitor all these results against targets the clinics have set for these different variables.
- The only ones are Title V, time to diagnosis and treatment (“all under care” is how I define it), report all presumptive positive results--I use the true positives, there are a lot of false positives, so I don’t use those, I use a percent of the true positives and how many of those got timely follow-up, referred and seen by the program,
- Title V reporting might be a specific task WSGSC could address.
- Time to treatment for NBS, no long term follow-up outcomes, for PKU and CF we have access to clinic data, we use data to evaluate our PKU and CF programs, once children are diagnosed and treated, the file goes to our CSHCN program, 45 conditions are tested for but we have only have support for PKU and CF in our CSHCN program.

Reimbursement

19. Has reimbursement for genetic services in your state improved, become worse, or stayed the same in the last year? If reimbursement has improved, in what ways has the collaborative contributed to the improvement?

Genetics

- Stayed the same. (3)
- Improved. Twice a year we (state agency) query which insurers accept CPT code 96040. There are more payors recognizing the genetic counseling code. The rate they pay is what the E&M coded as evaluation and management consultation codes. Consultation codes were mixed in before so we could not determine reimbursement. Physicians use another consultation code.
- The genetic services policy project looked at public and private payor for both lab and clinical findings. Prenatal and lab reimbursement were good. Pediatric cognitive services and medical genetic and genetic counseling and adult cognitive services are poorly reimbursed.
- Payors decline all genetic testing. Multiple literature reviews and Policy Project findings show variation in how people were doing things. Agency staff are talking with providers and billers to hear what they’re doing.
- We are still working on geneticist (not metabolic) orders. Esoteric tests are not reimbursed and we’re going to have to get preauthorization for practically everything anymore.
- Lab test coverage is abysmal. We’re working with insurers to figure out how to get preauthorization.
- Working with Medicaid for better reimbursement.
- Providence in our state writes off these tests.
- One lab in country does some of these tests and they don’t accept insurance, often are research based.
- Would like to collaborate on how other states are dealing with test reimbursement.
- We do not know because we have not formally analyzed it.
- If we didn’t all contribute our in-kind, it would be hard to make a go of it, it’s all mostly public money, geneticist need this support in funding from public programs.
- Endocrinology and hematology specialists are not getting reimbursed so they all go into general pediatrics to fund their practice. In our state, even for genetic services, we have a public-private partnership. We use our NBS to help fund genetics so we can have a geneticist.

Newborn Screening

- Both increased and decreased depending upon what it is:
  - Increased – we got funding from state to set-up a one time sickle cell collaboration. Also, we worked to get state funding to cover the gap in Medicaid coverage of treatment products.
Medicaid was insufficient and the state pays us to get treatment products to families who need them.

**Decreased** – with expansion of the NBS panel to fifteen conditions we have not had funding to expand our staff or the biochemical geneticist who respond to the positive screens. So because the numbers are up it’s a decrease in funding. The sickle cell physicians provide phone consultation that is not billable and what is billed is not fully reimbursed. Social work and nutrition counseling for these families is often not paid for either, so comprehensive services are not paid for.

- We cover our costs of NBS by the $60 billing fee. We supply metabolic food products for families who might have a hard time getting them. Because the conditions are rare, local pharmacies will not stock these. We can order in quantity and assure a reliable, dependable, inexpensive source for families in all areas, including rural.
- Insurance pays so little of medical specialty tests, confirmatory tests. Oregon helps us if families are uninsured, our program helps families to pay for testing, shipping and handling, coverage of confirmatory tests and specimen collection handling.
- Formulas, we’ve struggled with that, Medicaid doesn’t pay what they need to and suppliers have refused to take on Medicaid because they aren’t getting enough reimbursement for medical foods even though we have a law that mandates coverage for medical foods, it does not cover shipping and shipping for special food costs more than what the food costs. Shipping from mainland is expensive. No one is working on this.
- Areas that need change as far as NBS-reimbursement for specialty services as a whole. When we needed follow-up and DNA testing on probands, we paid for DNA testing.

**General Comment**

20. **Do you have comments or suggestions for the Collaborative that we have not covered during the interview?**

- Our dilemma is that our region is large and money is miniscule compared to geography and population, so we have a challenge to try and come up with useful projects beneficial for entire region.
- Top priorities if budget was not a problem-billing and reimbursement is a huge issue and gets to heart of access. If clinics can’t justify some level of income they won’t hire specialist, so there are fewer people to provide services.
- Cross border issues– We send personnel out of state, the more we send people out, the less we have here in our own state, long wait times, our state capacity is diluted by regional sharing.
- Current economic crisis makes me even more nervous about how people are going to get services.
- Health equity is a goal, then barrier is how are we paying for health care system, don’t have real information on access (we don’t know about access from family or other standpoints), how do people enter and navigate the health care delivery system and that’s where education comes in.
- Keep up the ability to bring everyone together to share information and ideas. Collaborative not directly helpful to me beyond information sharing (including hearing about national level information that I might not otherwise hear about), but is good for states that do not have programs and services.
- April NBS meeting was incredibly useful to bring metabolic “players” together for discussion.
- Work on increasing reimbursement for tests.
- Once again, feel like I’m beating a dead horse, but work on common ground for client satisfaction as a region.
- Did we ever get down to developing a minimum data set?
- Once a year get together is good to talk about everything.
- I did look at the menu of positive outcomes and think it looks great. I do think that some of this is collectible with the right indicators. Other outcomes I think would be difficult to measure (i.e. federal and state legislators use current info about genetics to write laws….)
• More specific initiative that gets concrete work done-strategy for how to effect reimbursement, we don’t get in to specific strategies to work for reimbursement.
• Outcomes project should be more helpful if we had more data to really evaluate what we’re doing.
• Who is going to read all this stuff, you almost do it as a requirement to – when ACMG came out with 29 disorder recommendation, a lot of NBS did something about this disparity. Now there’s a move for more than X disorders.
• It is all competing with people’s time. Getting worse and worse to try to get anything done, trying to make best of what you’ve got, but positions are vacant and you can’t go to conferences anymore, have to learn on your own, everything has gotten hard to do your job now, have competing priorities and all this (WSGSC work) is nice to have but unless there’s something tied to it people will not do.
• Would like to see our collaborative get more specific, we’re so general.
• I really like the NBS meeting because for first time I heard something specific, talked about cases, something more I could relate to, a specific outcome of people pooling data in our region and then getting a publication out of that was good.
• Do not know where we are with long term follow-up of NBS. Interesting to see how all states are going to implement guidelines that are coming out in the future.
• I wish we had in terms of our outcomes, some way to develop measures we could all use, compare across the board as a region how we’re stacking up.
• We hear a lot of stuff at meetings and we don’t think or hear about it ‘til next meeting. It’s not specific enough for me to say, great.
• Narrow the focus down rather than spread ourselves so thin – then we’d have something to show for it.
• CPT1 video for AK was excellent.
• PRODUCT with a capital P, each state should get a specific product. Something that will last long after the funding dries up, something that is sustainable, like telemedicine staying after money dries up. Anything that is here to stay after money dries up. Hope we use money wisely.
• Catalyst is WSGS – start it but keep it going.
• We should do a better job on keeping collaborators informed of our PI activities
• Possibly more sharing calls.
• The WSGSC spends too much time on NBS, this could be because of my affiliation with the NBS collaborative, what I know of genetics the conditions on NBS panel are a very small subset of genetics, NBS is almost exclusively metabolic. It is my opinion that if the WSGSC dealt with NBS maybe 5% of its time and spent more time on other conditions, I might find more value there because I’m not getting that information somewhere else.
• People in WSGSC are NBS people so they emphasize that. Grossly disproportional amount of time on NBS.
• Too much time on reimbursement.
• Will not be coming to Summit.

References