

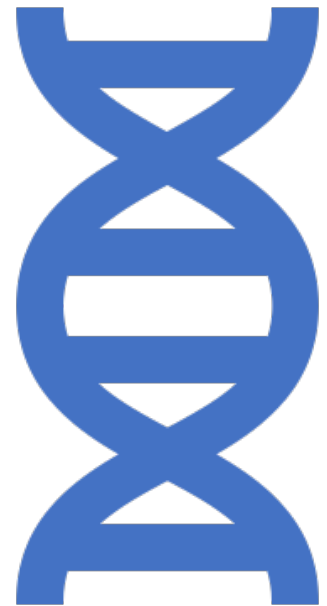
We finally got a
diagnosis!

It's rare.

What do we do now?

Greta L. Goto, MBA

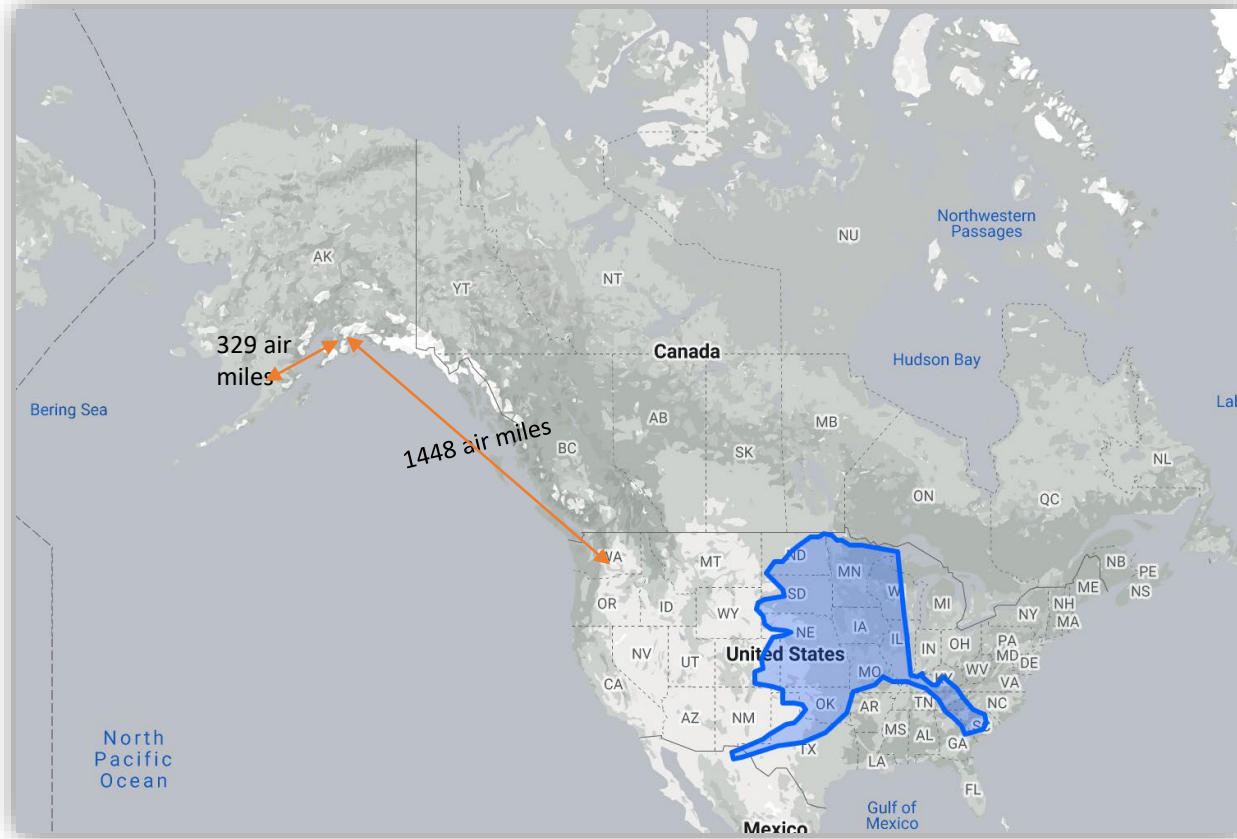
October 12, 2022





0-4 yrs
Failure to
thrive.....

Our diagnostic journey...



- started in Dillingham, AK
- to Anchorage, AK
.....and back and forth
 - *avg. roundtrip airfare \$450-600*
- and then, with a travelling genetics clinic from Seattle Children's Hospital, WA

Map source: The True Size

[https://www.thetruesize.com/#?borders=1~!MTxNTA0Mzc.NTExMDQwOQ*MTY3NjgyOTI\(MzA1MTQzODY~!CONTIGUOUS_US*MTAwMjQwNzU.MjUwMjM1MTc\(MTc1MA~!IN*NTI2NDA1MQ.Nzg2MzQyMQ\)Mg~!CN*OTkyMTY5Nw.NzMxNDcwNQ\(MjI1\)MQ~!US-AK*NDQ2NDc5MA.Mjc3NDQ1MjM\)Mw](https://www.thetruesize.com/#?borders=1~!MTxNTA0Mzc.NTExMDQwOQ*MTY3NjgyOTI(MzA1MTQzODY~!CONTIGUOUS_US*MTAwMjQwNzU.MjUwMjM1MTc(MTc1MA~!IN*NTI2NDA1MQ.Nzg2MzQyMQ)Mg~!CN*OTkyMTY5Nw.NzMxNDcwNQ(MjI1)MQ~!US-AK*NDQ2NDc5MA.Mjc3NDQ1MjM)Mw)



0-4 yrs
**Failure to
thrive.....**



Elementary
**Finally a
diagnosis at age
4.5 yrs**



0-4 yrs
Failure to thrive



Elementary
**Finally a
diagnosis at age
4.5 yrs**



**Middle & High
school**
**Serious
challenges and
need for
appropriate,
safe and trained
caregivers for
her continued
development
and
independence**

Applying for a Medicaid Waiver in AK

- Gather documents
- Get help from the aging and disability resource center to find appropriate resources
- Find and select a care coordinator
- Complete an intake packet with all relevant information about disability
- Apply for Medicaid
- Phone interviews, have additional documentation at the ready
- Nursing home level of care assessment
- If eligible, determine next steps of what resources needed for care
- Wait
- Keep care coordinator informed of any changes
- Develop plan of care
- Make sure supports are available and can take you and know your care needs
- Wait for approval of plan of care
- Implement plan of care
 - For renewals, complete Medicaid renewal
 - Complete level of care
 - Complete plan of care



0-4 yrs
**Failure to
thrive.....**



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**Middle & High
school**
**Serious
challenges and
need for care to
keep her safe w/
opportunities
for continued
development
and
independence...**



Age 22
**More
Transitions and
the Disability
Cliff.....**



0-4 yrs
**Failure to
thrive.....**



Elementary
**Finally a
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**Middle & High
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**Serious
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Age 22
**More
Transitions and
the Disability
Cliff.....**



And beyond...
**Aging and
related issues
for her and
parents and
caregivers**

Services used or needed over her life span....doesn't include family/sibling impacts/needs

Ayanna

- “I want to be with you....I want to stay here.”
- “I want to work.”
- “They’re not listening to me.”
- “Does that person support disabilities?”







Our observations

- Long journey to diagnosis—has ripple effects
- Scarcity of services & resources for genetic and rare disorders, especially in rural states and regions
 - *Alaska: have to fly to urban hubs; few resources outside "Railbelt"; few genetic counselors; genetics providers come up from "lower 48"; shortage of care coordinators; no medical school*
 - *What about the US Commonwealths and Territories?*
- No easy process to access services and supports outside Alaska
- Difficult to find support systems
- Transitions are hard— daily, school, medical, services, age
- Little confidence in health insurance providers understanding the complexity of a genetic disorder and needs for ongoing care
- Parents become the experts, yet they are not often respected as such
- Parental burnout, sibling supports needed, extended family provides care too

Alaska specific ideas to improve access and availability of genetic and rare care resources

- Ensure access to Genetic and Genomic testing, counseling and related resources
- Rare Informed Research, Education & Training
 - Institutionalize research and reporting on Alaska Genetic and Rare Disorders
 - Genetic and Rare Care Services mapping – current/future needs
 - Support access to rare patient registries and clinical trials
 - Develop virtual specialty clinics for diagnosis, treatment and management
 - Develop continuing education, training, and workforce development programs specific to genetics, genomics and rare disorders
- Conduct needs assessment with known rare disorder population AND their caregivers
- Research and Implement integrated care coordination & family supports & services inclusive of both medical and social needs for persons living with genetic and rare disorders and their caregivers
- Alaska Genetic and Rare Disease Community Advisory Council
 - Genetic and Rare Care Advocacy, Partnerships, Fundraising



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