

National Prion Disease Pathology Surveillance Center

2025 Update & Panel



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Summary

- Provide a brief overview of the NPDPS
- Provide some basic info on our findings
- Q&A Panel with NPDPS faculty and staff



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NPDPS Mission

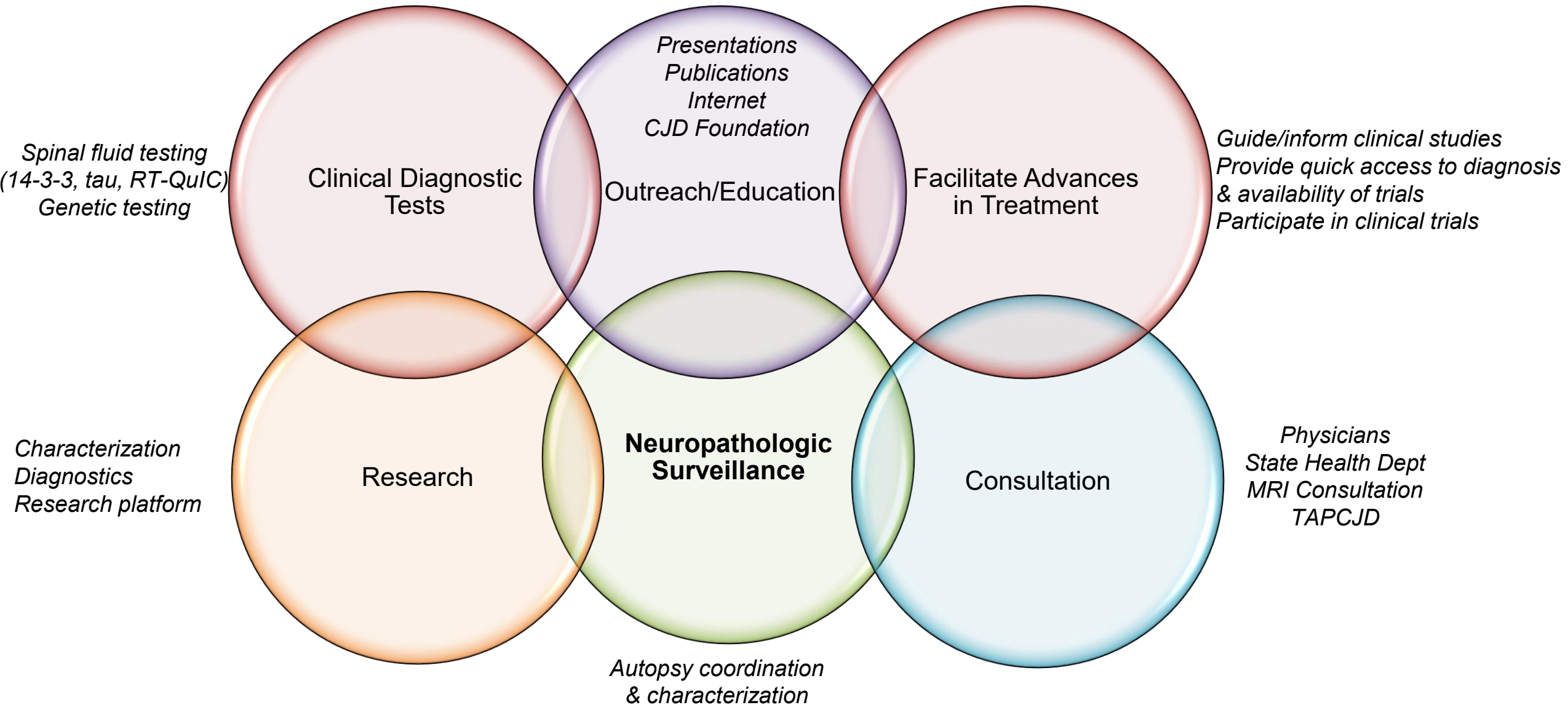
- Started in 1997 at Case Western Reserve University in response to variant CJD by CDC in collaboration with Dr. Pierluigi Gambetti
- Neuropathologic surveillance activities are funded by CDC
 - Allows for a definitive diagnosis of prion disease
 - Determines the type of prion disease
 - Allows for neuropathologic and biochemical surveillance for novel forms of the disease
- Important focus is on preventable & potentially novel forms of prion disease (e.g., variant CJD, iatrogenic CJD, and novel acquired prion diseases)



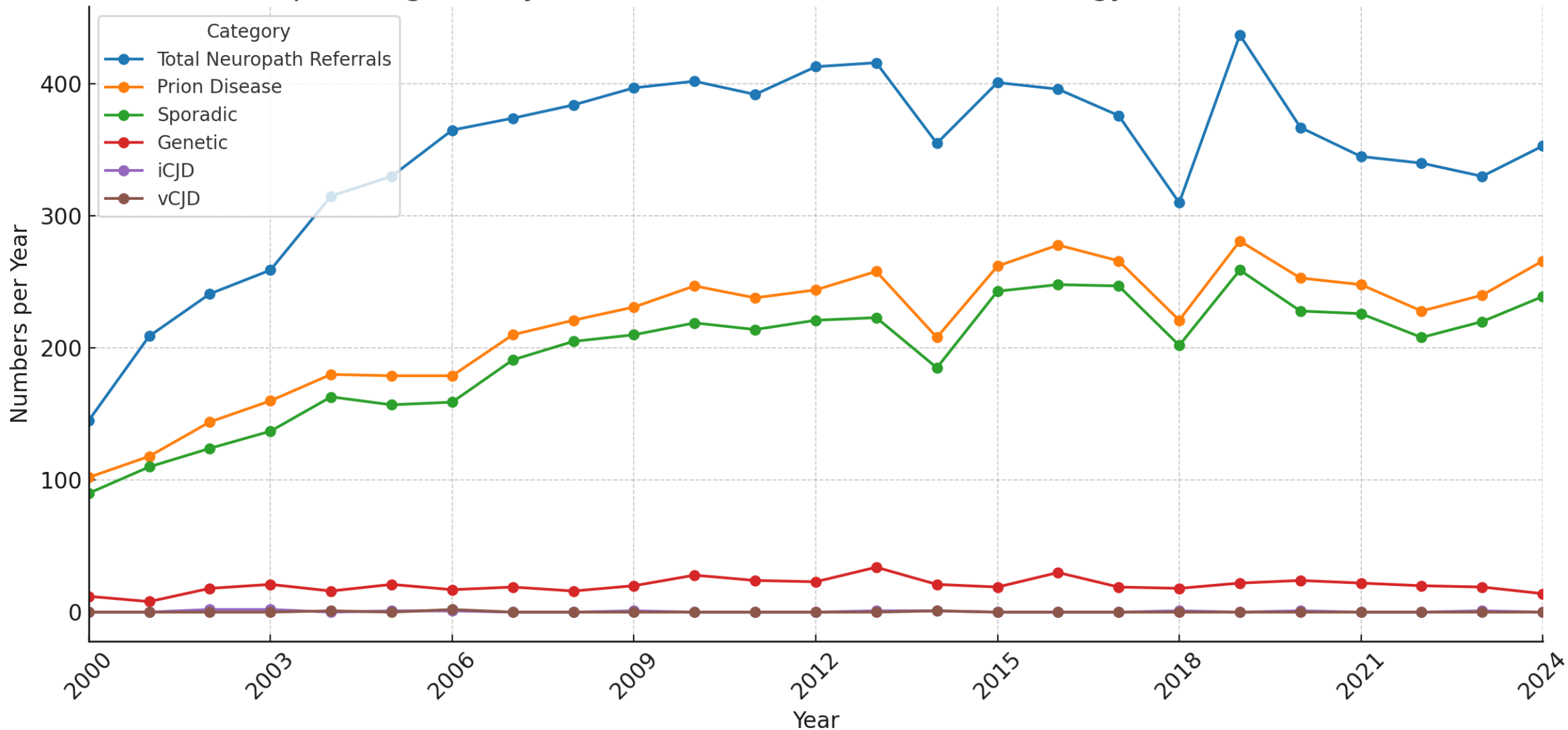
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Aspects of the NPDPS

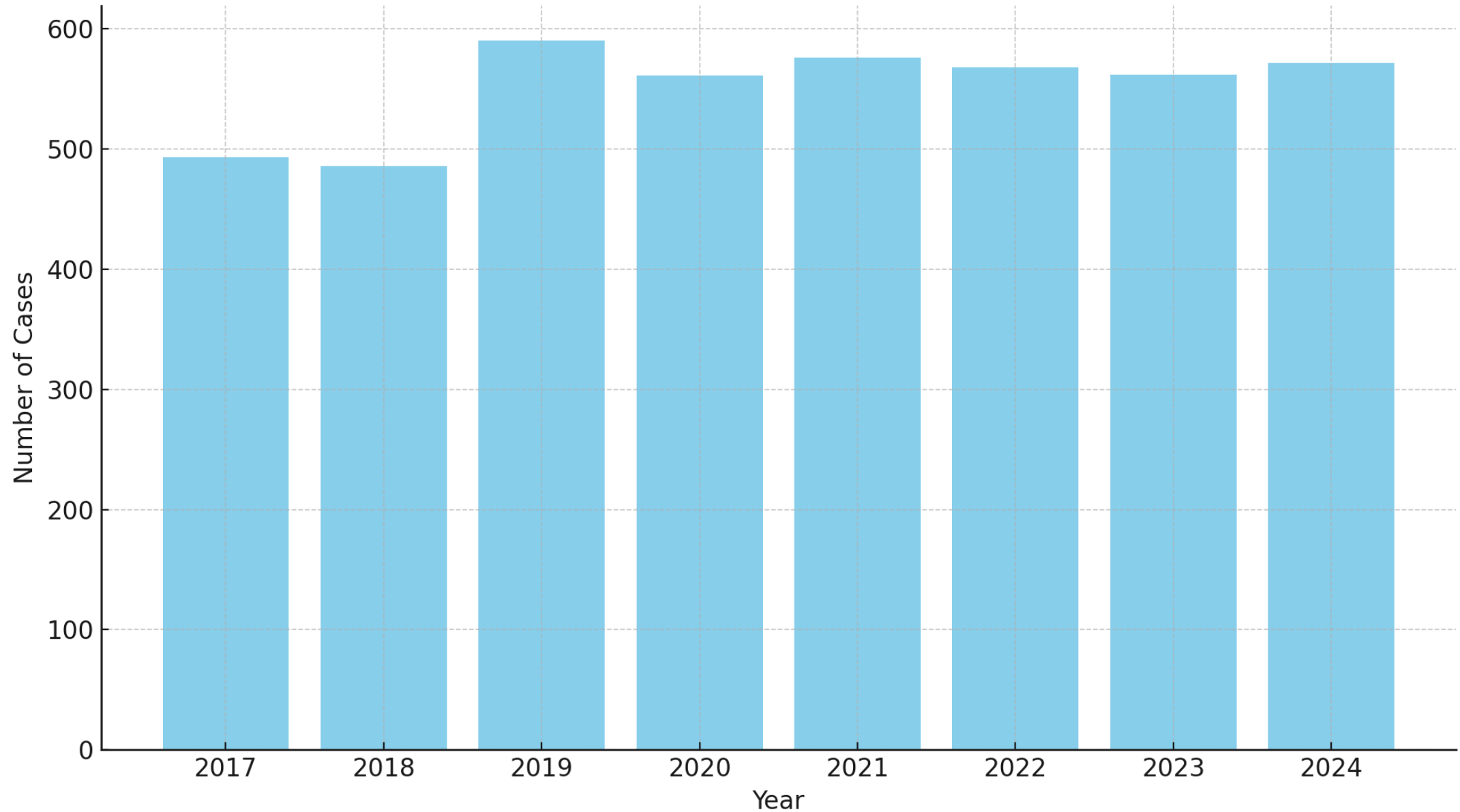


Neuropathologic Analyses at National Prion Disease Pathology Surveillance Center



vCJD: 4 cases (all exposed outside the US)
iCJD: 15 cases, mostly from c-hGH

Probable & Definite Prion Disease Cases by Year



Definite = Autopsy confirmed
Probable = Autopsy confirmed + Positive RT-QuIC cases that did not go to autopsy

Sporadic Prion Disease Distribution in the US

Sporadic Subtype	2024 %
MM1	36
MM1-2	15
MM2	4
MV1	2
MV1-2	13
MV2	11
sFI	0
VPSPr	1
VV1	1
VV1-2	8
VV2	8



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Genetic Prion Disease Mutations in the US

Mutation	Total	Mutation	Total
E200K-129M	322	V203I-129M	3
Insertions	96	V180I-129V	3
V210I-129M	88	P102L-129V	2
D178N-129M	69	Q217R-129V	2
E200K-129V	57	H187R-129V	2
P102L-129M	55	E196K-129M	2
D178N-129V	37	A224V-129V	1
T188R-129V	29	G131E-129M	1
A117V-129V	24	G94S-129V	1
H187R-129M	10	H187A-129M	1
F198S-129V	10	N171S-129V	1
V180I-129M	10	Q160X-129M	1
R208H-129M	6	Q186X-129V	1
P105S-129V	5	R148H-129V	1
G54S-129M	5	S245P-129M	1
V203I-129V	4	T183A-129V	1
T183A-129M	4	T188A-129V	1
R148H-129M	4	T188K-129V	1
E200G-129V	3	V189I-129M	1
E196A-129M	3	P105S*	1
A133V-129M	3	Deletion	1
G114V-129M	3		
R208H-129V	3		
Subdivision of Insertions			
Mutation	Total	Mutation	Total
6 Inserts-129M	21	9 Inserts-129V	5
5 Inserts-129V	20	1 Insert-129M	3
5 Inserts-129M	14	9 Inserts-129M	1
4 Inserts-129M	10	1 Insert-129V	1
6 Inserts-129V	10	8 Inserts-129M	1
2 Inserts-129M	9	8 Inserts*	1
Subdivision of Deletions			
2 Deletions-129M	1		
Cases could not be haplotyped (P105S, 8 OPR1)			



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Huge Thank You!

- CJD Foundation
- Akin
- Families
- Friends (fellow scientists/collaborators)



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NPDPSA Panel

- Katie Glisic, MA: Admin Lead
- Meghan Lewis, MA: Autopsy Program Manager
- Eriton Cunha, PhD: Protein Chemistry
- NPDPSA staff in the audience
- You!



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