

The ATHNdataset: Rare Coagulation Disorders

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Working Group: Thank you!

PAI-1, Plasminogen & HHT

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Working Group: Thank you!

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Vitamin K Dependent Proteins, Factors V, V & VIII, X

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Goals

- How do we collect meaningful data going forward to
 - Inform unanswered questions
 - Improve our understanding of, and patient outcome for these rare disorders
- What data capture capability we now have within WebTracker and how this guides our future processes

ATHN desires to support national and international efforts to increase visibility and advocacy for rare coagulation disorders

Our story:

Rare Coagulation Disorders

- Rare disorders
 - Affects 1:7500 – 1:2,000,000 patients
 - Treatments limited
 - Research requires global population & collaboration
- Challenges for data collection
 - Disease specific meaningful data
 - Patients within and outside of HTC network
 - Compatibility with international databases
 - European Network on Rare Bleeding Disorders

Rare Inherited Hemorrhagic & Thrombotic Disorders

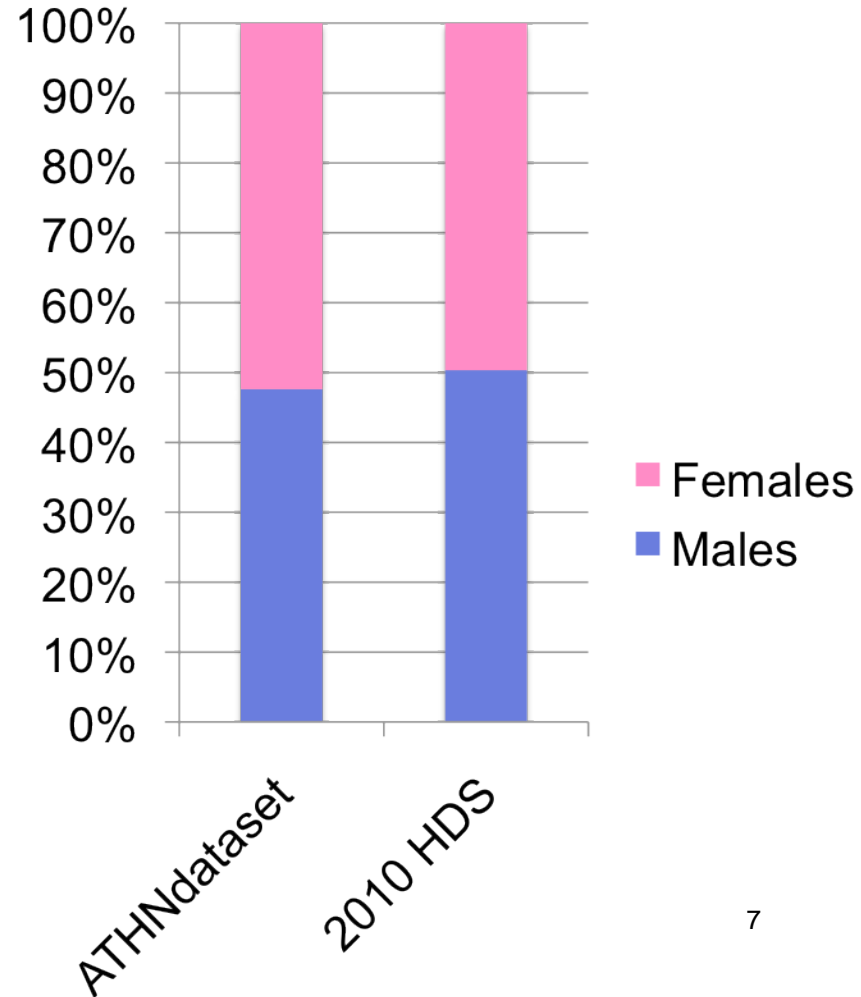
Clotting Factor / Disorder	Clinical Phenotype	
	Hemorrhagic	Thrombotic
Single Deficiency		
Fibrinogen (I)	✓	✓
Factors II, VII, X, V, XIII, XI	✓	
PAI-1	✓	
α 2antiplasmin	✓	
Protein C, S, AT3		✓
Plasminogen		± ✓
Combined Deficiency		
FV & VIII	✓	
FII, VII, IX, X	✓	
Miscellaneous Disorders		
APLS, HHT, Ehlers Danlos	✓	✓

Rare Bleeding Disorder Patient Gender

ATHNdataset (as of September 1, 2011) vs. 2010 HDS

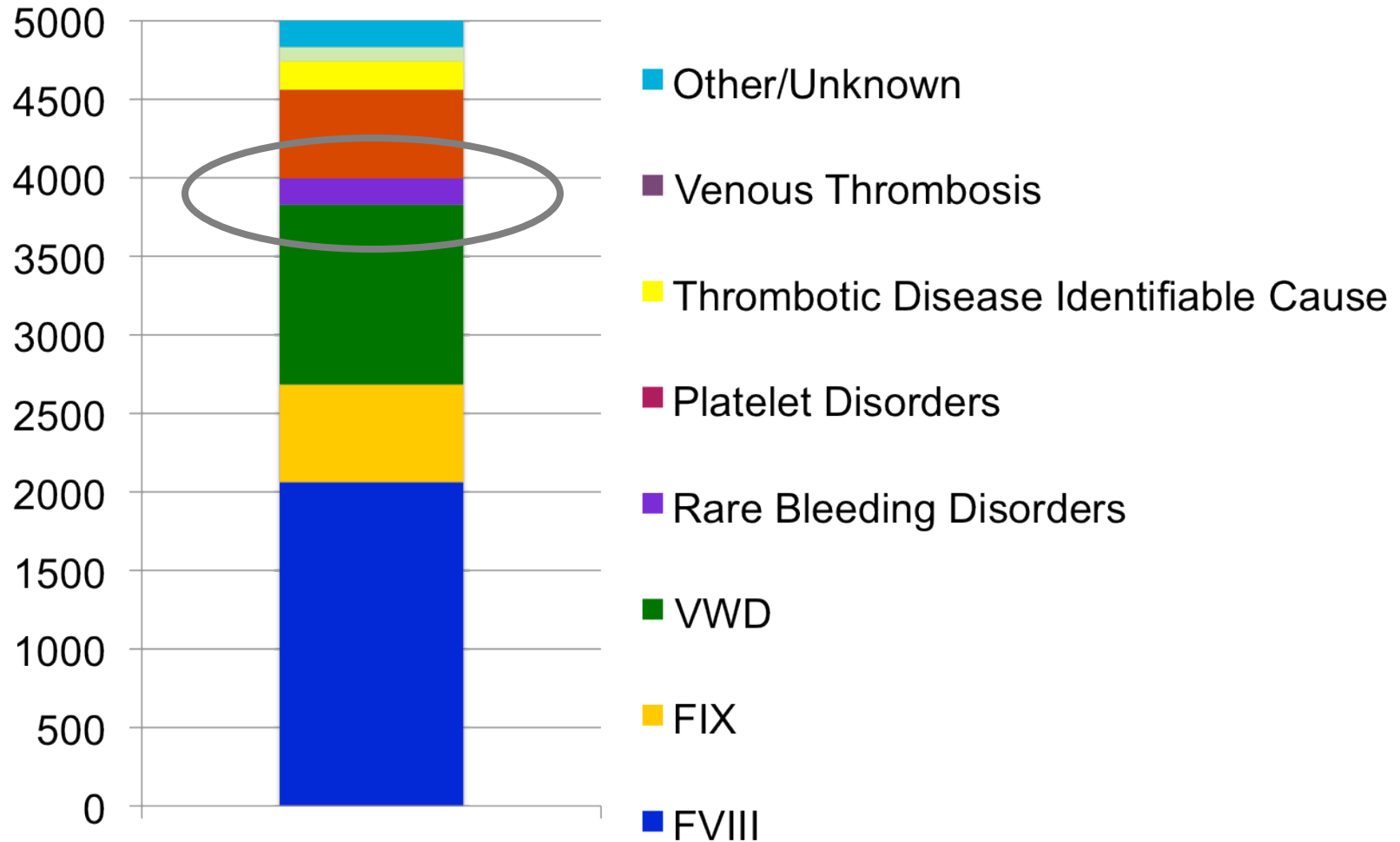
ATHN	RBD Patients # (%)
Male	70 (48%)
Female	77 (52%)
Total	130

HDS	RBD Patients # (%)
Male	892 (50%)
Female	880 (50%)
Total	1772



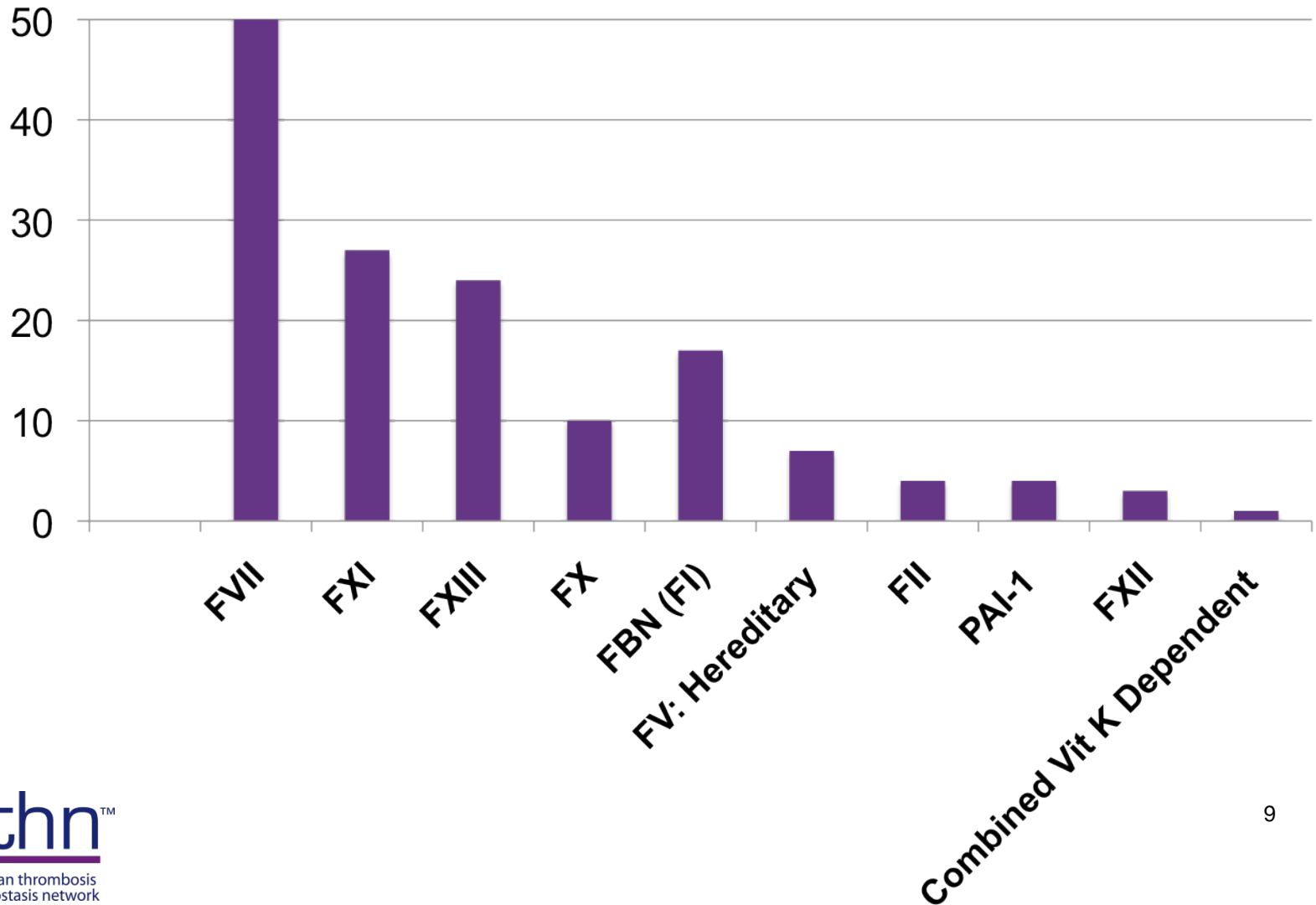
Patients By Primary Diagnosis

(5,141 Authorized Patients as of September 1, 2011)



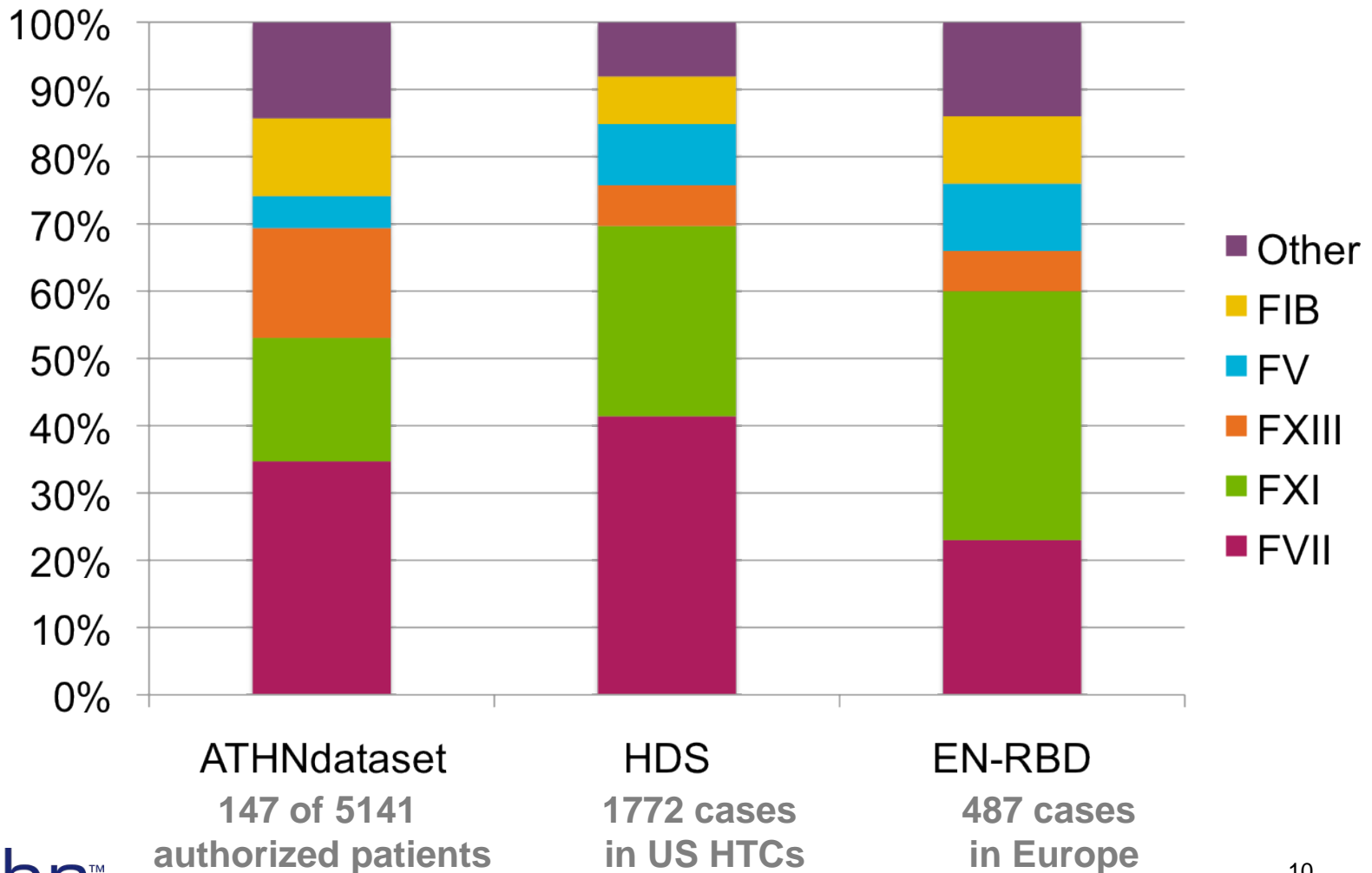
Rare Bleeding Disorders Patients

(5,141 Authorized Patients as of September 1, 2011;
147 Rare Bleeding Disorders Cases)

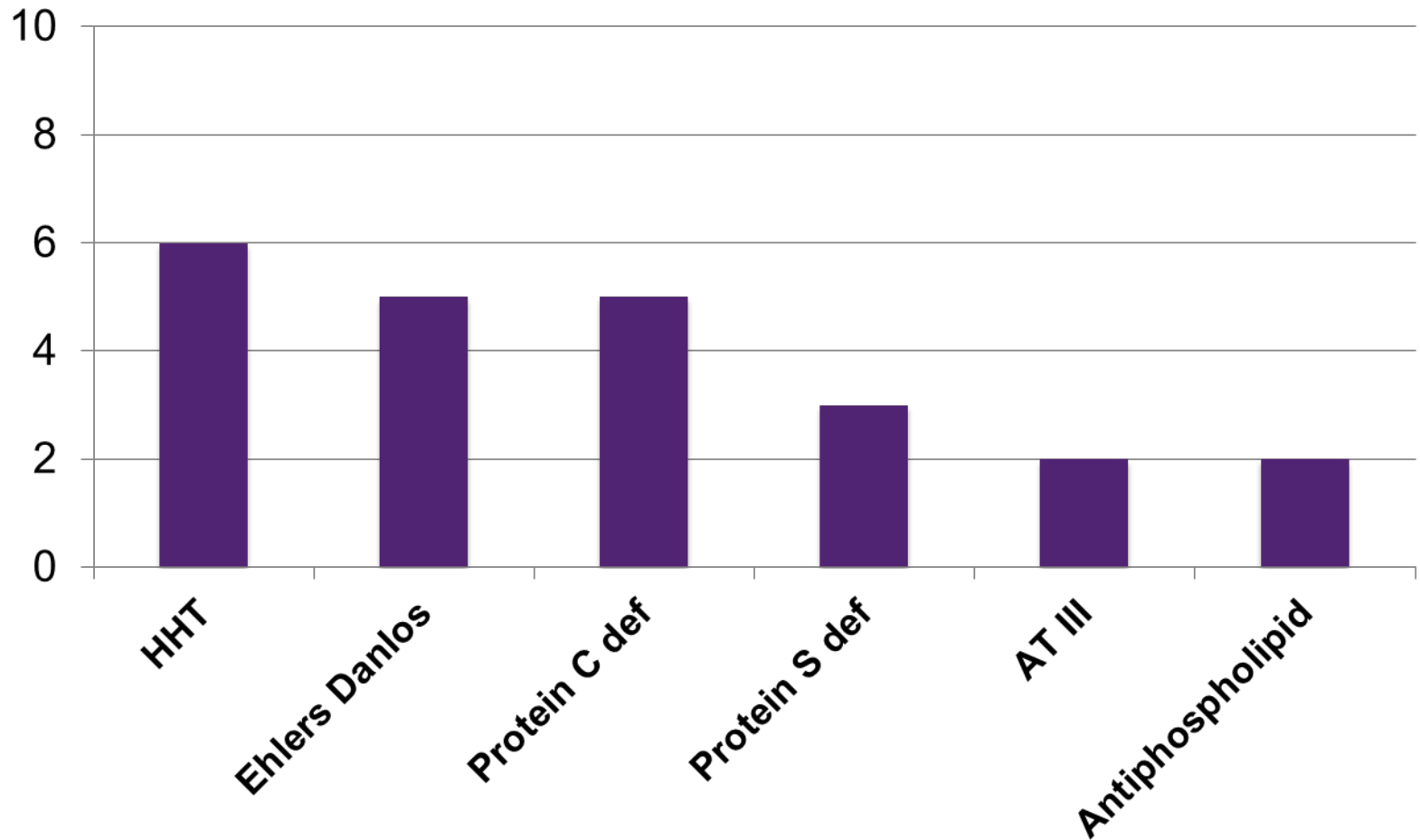


Rare Bleeding Disorder Patients

ATHNdataset as of 09-1-2011 vs. 2010 HDS and EN-RBD

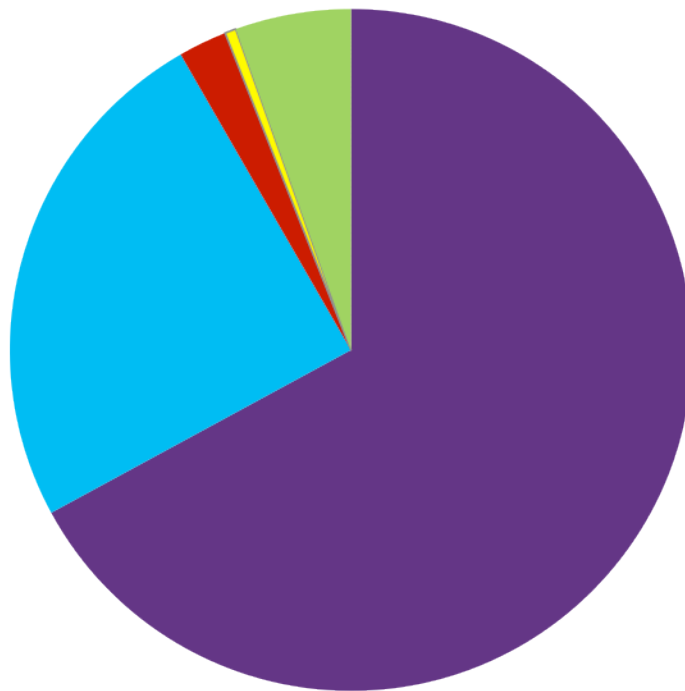


Other Rare Disorders in ATHNdataset (5,141 Authorized Patients as of September 1, 2011; 23 Cases)



Platelet Disorders Patients

(5,141 Authorized Patients as of September 1, 2011;
565 Platelet Disorders Cases)



- Storage Pool Disease
- Platelet Function Disorder Unspecified
- Glanzmann Thrombasthenia
- Bernard Soulier Syndrome
- Other

Relevant Data Elements Available Through WebTracker & ATHNadvoy

DEMOGRAPHICS	CLINICAL INFO	OUTCOMES
Date of birth	Primary/other diagnoses	Bleed events
Race	Date of diagnosis	Bleed location
Ethnicity	Disease severity	Prophylaxis used
Gender	Factor levels	Treatments used
Age at 1 st HTC visit	Treatments prescribed	Date of death
Marital status	Age at 1 st exposure	Cause of death
Education level	Surgeries/procedures	Anemia
Contacts	Inhibitor status	
Languages	Allergies	
Payer type	Molecular defect	
	Phenotypic studies	

Summary

- Proof of concept confirmed
 - Rare disorders can be captured using WebTracker structure
- Data collection has started and is included in ATHNdataset
 - Target 100% of rare coagulation disorder patients within HTCs to be registered in 2012
- Issues remain
 - Data capture on patients outside HTC network
 - Additional data elements of interest such as reproductive history yet to be incorporated