Clinical symptoms in patients suspicious to lysosomal storage disorders in Germany, Austria and Switzerland

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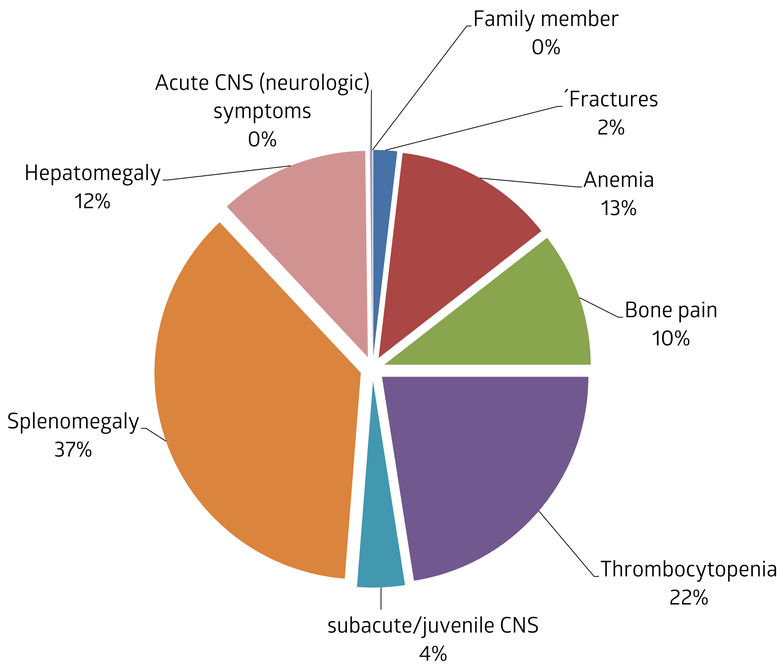
Introduction:

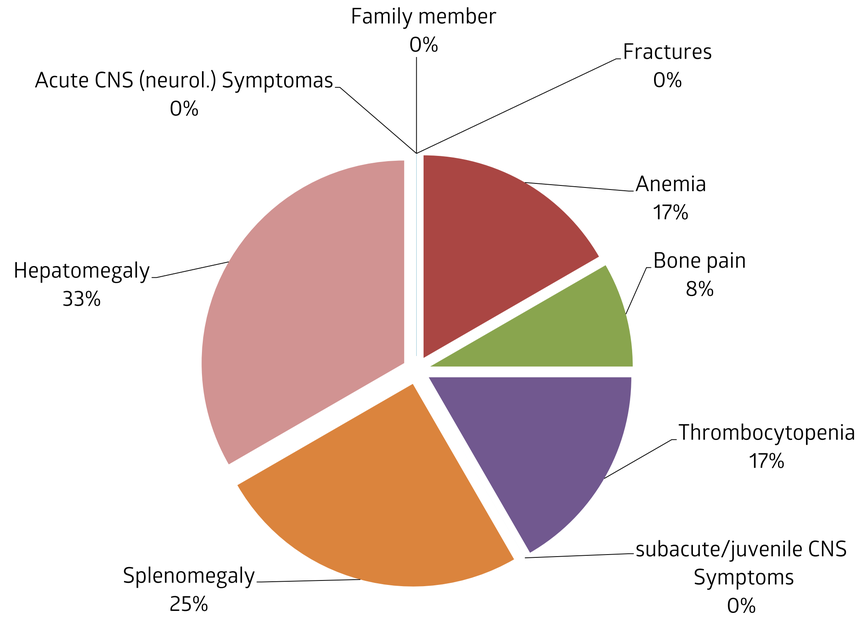
We present our experience and data in providing a comprehensive diagnostic initiative in Germany, Austria and Switzerland started in 2013. In those countries no newborn screening for any LSD exists thus identifying any person suspicious to LSD disease by primary diagnostic testing often occurs when symptoms are already present. Consequently, often final diagnosis is delayed with negative impact on therapy management and clinical outcome.

Gaucher disease:

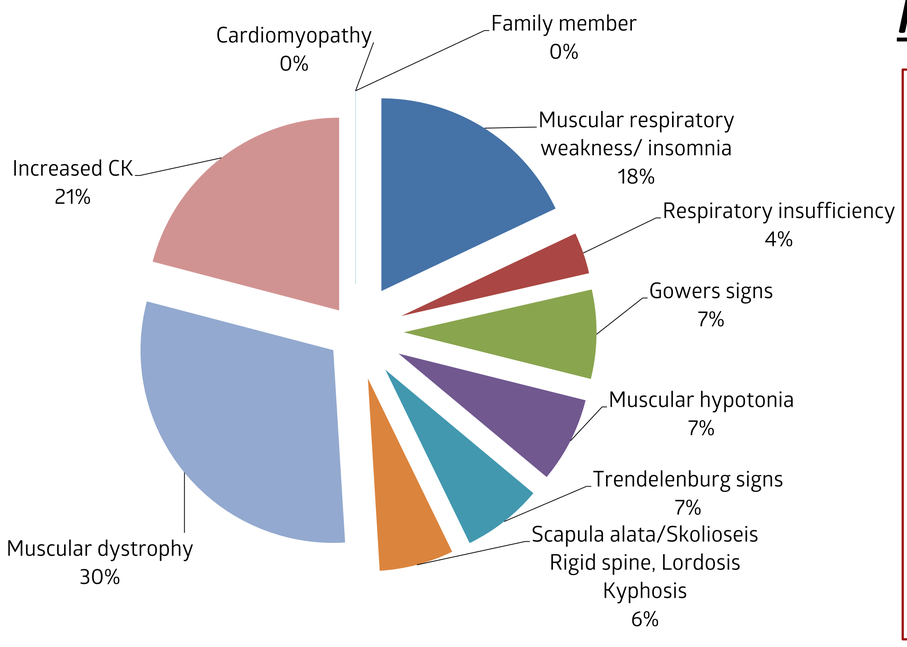


Percentage of related symptoms of 262 tested persons

Percentage of symptoms of genetic confirmed (8 of 262)

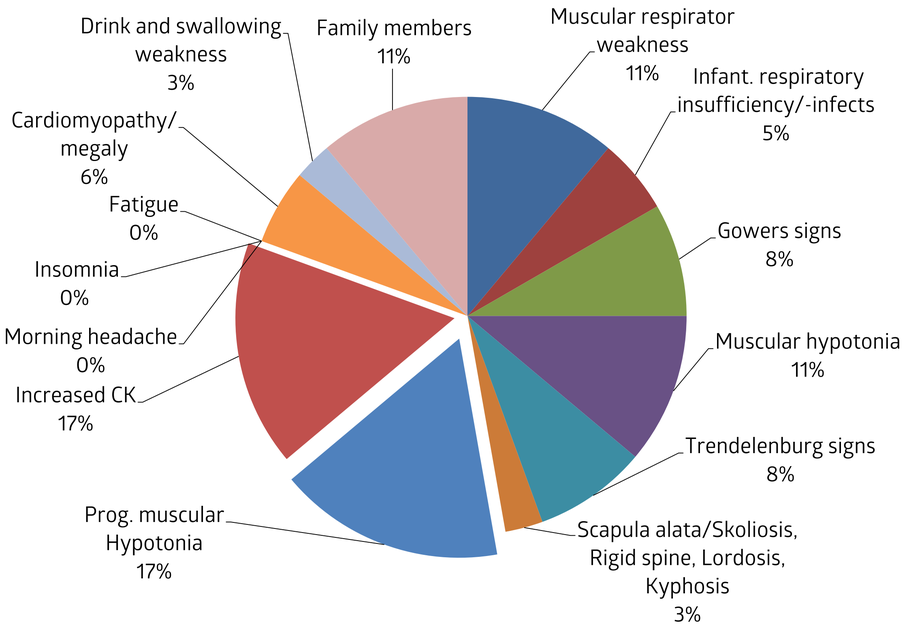


Pompe disease:

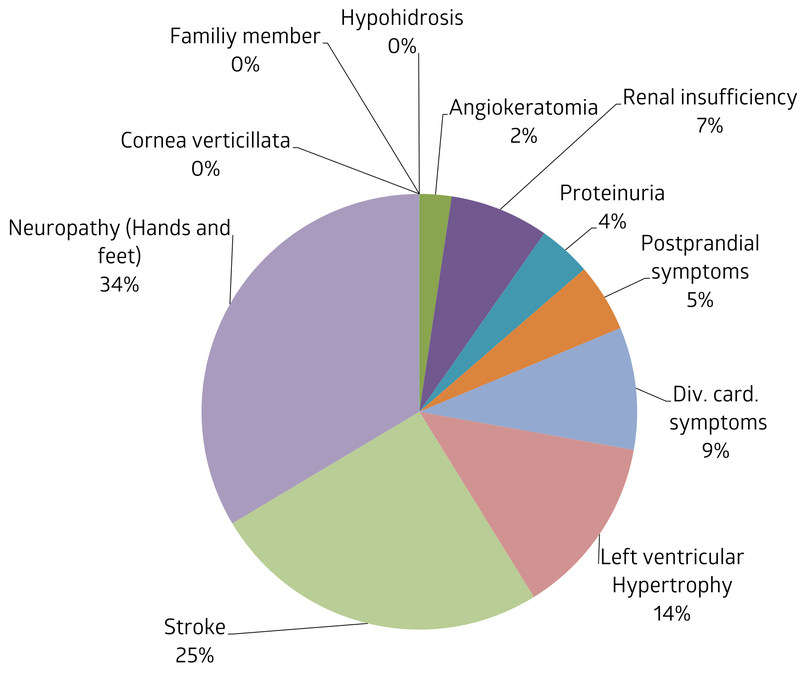


Percentage of related symptoms of 749 tested persons

Percentage of symptoms of genetic confirmed (20 of 749)

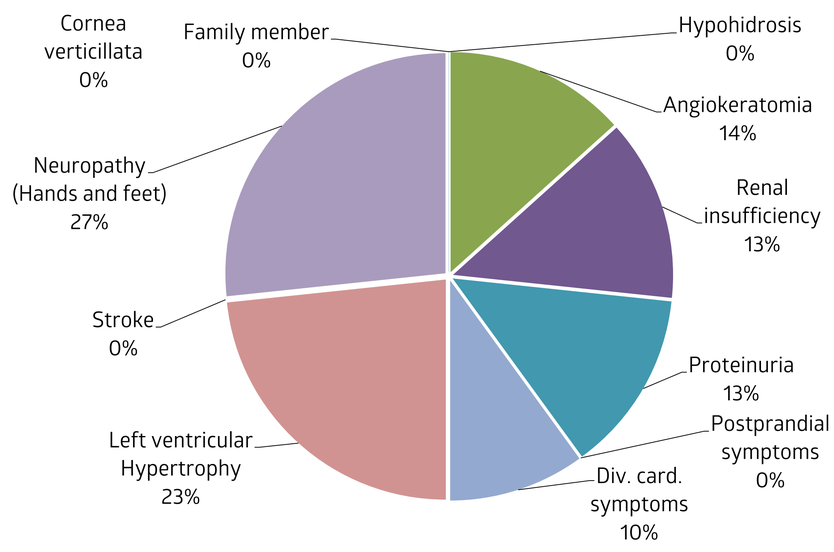


Fabry disease:



Percentage of related symptoms of 1789 tested persons

Percentage of symptoms of genetic confirmed (19 of 1789)



Conclusion:

The combination of a disease awareness campaign as well as providing simple, fast and high quality diagnostic using Dried Blood Spot testing for enzyme activity, genetic as well as biomarker testing, dozens of newly diagnosed patients could be identified. In addition, clinical signs at time point of diagnostic testing were collected. These data are shown in comparison to those symptoms found in genetically confirmed patients. With these data, we hope to better characterize patients and their clinical presentation at doctor's visit and to provide in future physicians a more specific clinical picture of patients with LSDs. The diagnostic service is supported by Genzyme Germany.

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