



Results of the Hamburg pilot study for newborn screening of hemoglobinopathias

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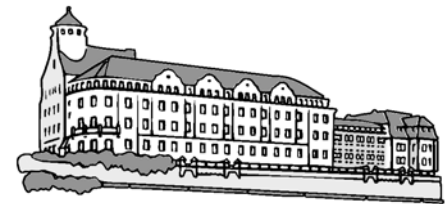
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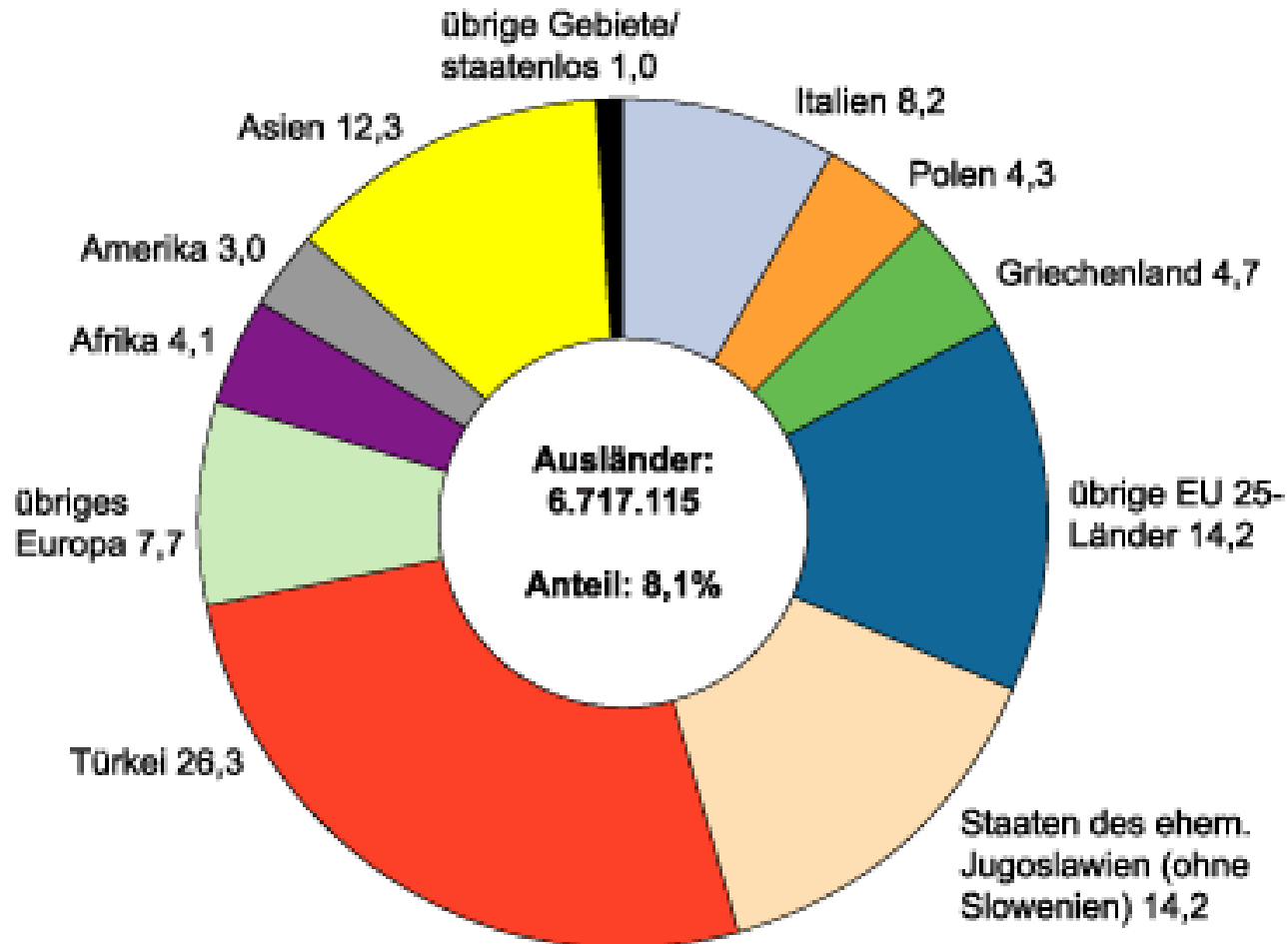
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FÜR TROPENMEDIZIN

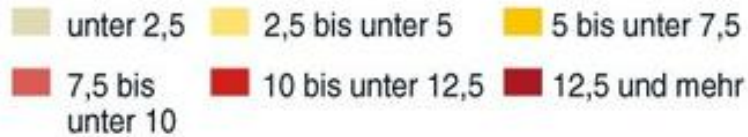
Geographical Origin of foreigners who live in Germany

Staatsangehörigkeiten mit einem Anteil von über 4% sind einzeln aufgeführt, die anderen nach EG/EU bzw. Kontinenten zusammengefasst.



Foreigners in Germany

Anteil der Ausländerinnen und Ausländer in Prozent in Regierungsbezirken und Ländern 2009



Schleswig-Holstein

5,1 %

Hamburg

14,0 %

Bremen

12,7 %

Niedersachsen

6,8 %

Nordrhein-Westfalen

10,5 %

Hessen

11,1 %

Rheinland-Pfalz

7,6 %

Saarland

8,4 %

Baden-Württemberg

11,8 %

Mecklenburg-Vorpommern

2,3 %

Brandenburg

2,6 %

Berlin

13,8 %

Sachsen-Anhalt

2,7 %

Sachsen

2,7 %

Thüringen

2,1 %

Bayern

9,3 %

Deutschland

8,7 %

Quelle: Destatis

AFP

Historical Data concerning hemoglobinopathias in Germany

- There is no comprehensive data concerning sickle cell disease or hemoglobinopathias in Germany
- Data from the Ulm reference laboratory:
1971 – 2007: 100.621 hemoglobin analysis,
3085 persons identified with sickle cell disease
(Kohne et al, Dtsch Arzteblatt 2011)

Goals of hemoglobinopathy screening



- lower morbidity and mortality by early prevention measures and by this, lower costs for sickle cell treatment.
- Improvement of therapy options and better information of affected couples

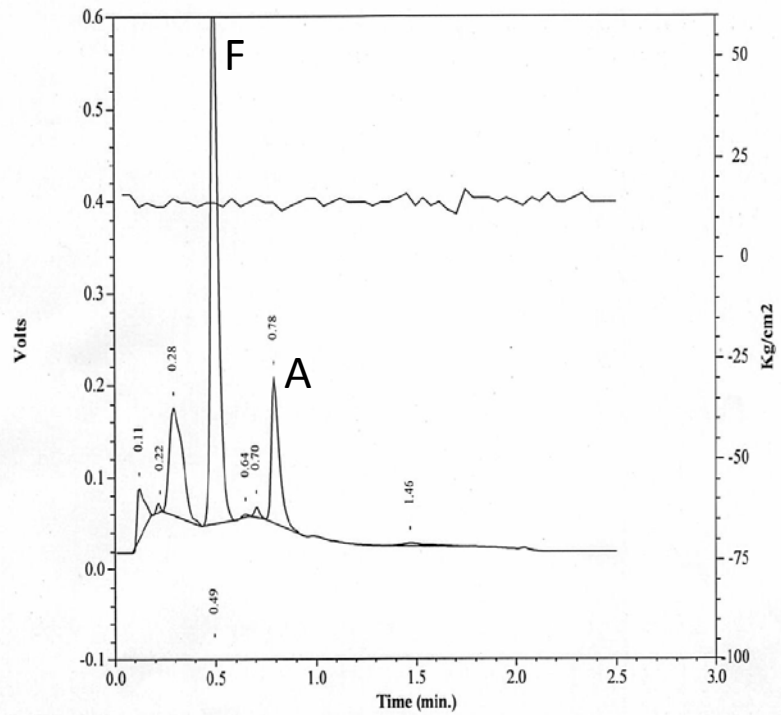
Method



Chromatograms



Pattern: FA
Total Area: 2691830





Screening Study

- 17 018 newborns have been included in the study
- all children born in the greater Hamburg area
- no bias
- anonymized samples
- 321 samples have been excluded because of poor sample quality
- First-tier testing : HPLC-analysis
- second-tier testing : molecular genetic testing (same sample)



Results

- **FAST > 10% referred to genetic testing for α -thalassemia**
- **Hb-E/A2-peak > 3.5% referred to genetic testing for β -thalassemia**
- **HbS > 5% referred to genetic testing for SCD**

on SCD and SCT Duffy blood group testing was carried out as well



Results

TABLE I. Results of HPLC and Molecular Analysis

Hemoglobin pattern (HPLC)	<i>HBB</i> Genotype	<i>DARC</i> promotor Genotype	Newborns (n)
HbF, HbS	<i>HBB</i> : c.20A>T/c.20A>T	C/C	3
HbF, HbS, HbC	<i>HBB</i> : c.20A>T/c.19G>A	C/C	4
	no material		1
HbF, HbA, HbS	<i>HBB</i> : c.20A>T/wt	C/C	59
	<i>HBB</i> : c.20A>T/wt	C/T	20
	<i>HBB</i> : c.20A>T/wt	T/T	9
	<i>HBB</i> : c.327 C>G (Hb Presbyterian)/wt	C/C	1
	<i>HBB</i> : wt/wt	T/T	3
	no material		6
HbF, HbC	<i>HBB</i> : c.19G>A/c.19G>A	C/C	1
	no material		1
HbF, HbA, HbC	<i>HBB</i> : c.19G>A/wt	C/C	22
	<i>HBB</i> : c.19G>A/wt	C/T	5
	<i>HBB</i> : c.19G>A/wt	T/T	1
	<i>HBB</i> : wt/wt	T/T	1
	no material		4
HbF, HbA, HbE	<i>HBB</i> : c.79C>T/wt	T/T	7
	no material		4
HbF, HbA, HbD	<i>HBB</i> : c.364G>C/wt	T/T	11
Total			163

Results



	Berlin <i>Lobitz et al, 2014</i>	Hamburg <i>Grosse et al, 2016</i>	Baden- Württemberg <i>Kunz et al, 2016</i>
Heterozygote HbS	1 / 129	1 / 189	1 / 456
SCD	1 / 2.435	1 / 2385	1 / 12.000 (estimated)
Total samples	34.084	16.697	37.838

SCD was the most prevalent disease in newborn screening of Hamburg and Berlin

Summary



- reliable prevalence data is now available
- It was shown that screening for SCD is effective and necessary in Germany
- Recent immigration will further increase the prevalence
- Political decision required for selective or whole-population screening
- Prerequisite: Improve structures for counseling and treatment

time
is
NOW.



Thank you very much!

