Clinical Genetics in China: Genetic Disorders and Current Status

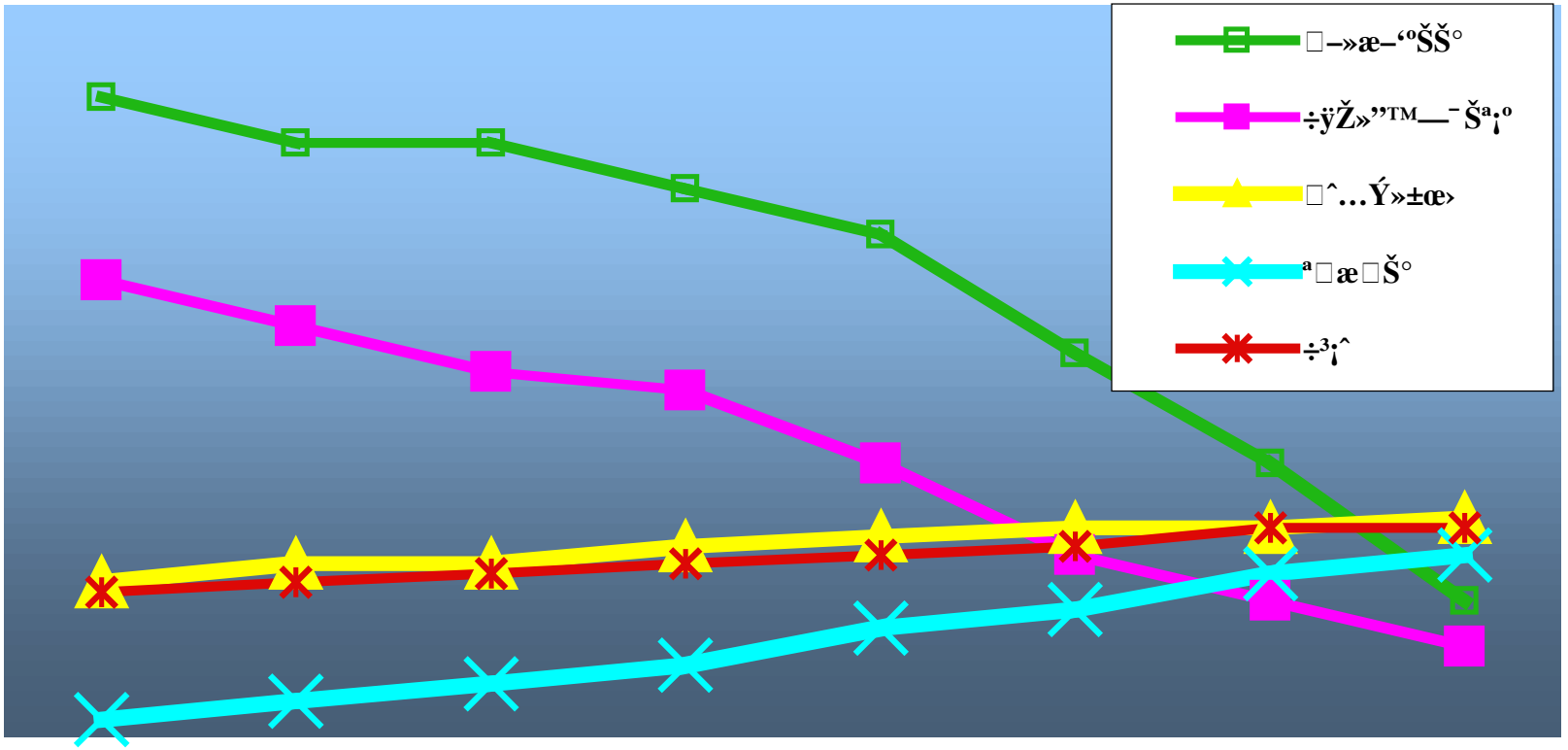
Content

- Clinical Genetics
- Infrastructure of national public health
- Policy and regulation
- Genetic testing network
- Capability of genetic testing
- Genetic screening
- Translational medicine

Present situation of clinic genetics

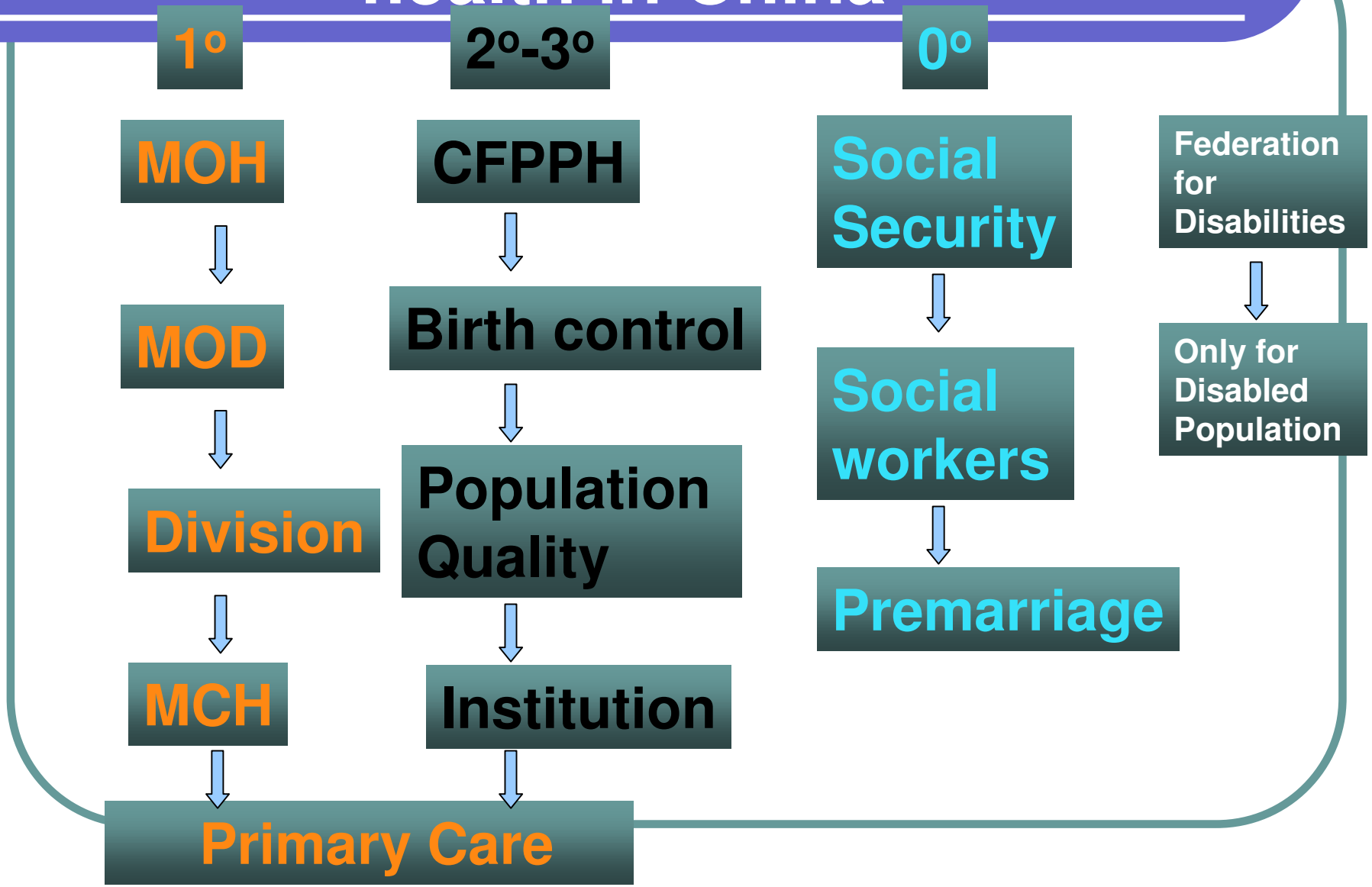
- Birth Defects：4-6‰
- Tri-level network：1st^{o}, 2nd^{o}, 3rd^{o}
  - Large population
  - Broad landscape
  - Imbalanced development
- Undeveloped translational medicine
- Coverage
  - Most of the testing is carried out by academic laboratories
  - Commercial testing is focusing on “risk assessment”

Spectrum of Newborn Disorders in the last 30 years



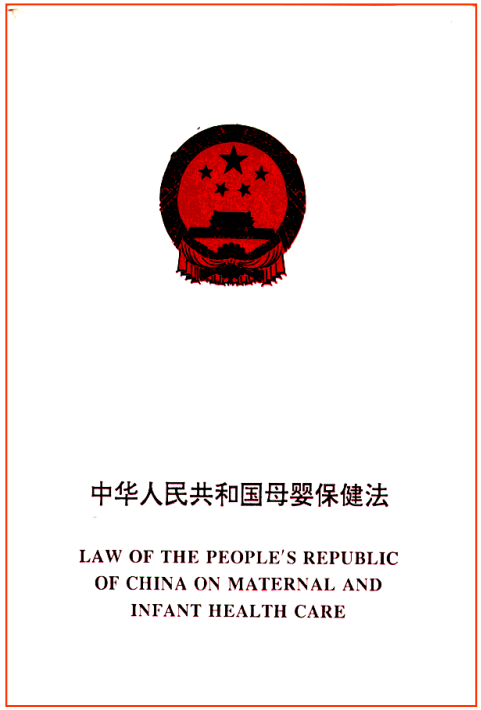
From: UNICEF 2000 Annual Report

National Infrastructure for Public health in China



Policy and Regulation

- 1994：**National Law**“对婴儿开展预防接种（vaccine)，逐步开展新生儿疾病筛查(neonatal screening)”
- 2002：卫生部、残联《中国提高出生人口素质，减少出生缺陷和残疾行动计划(2002-2010）》
- 2009:卫生部《新生儿疾病筛查管理办法》



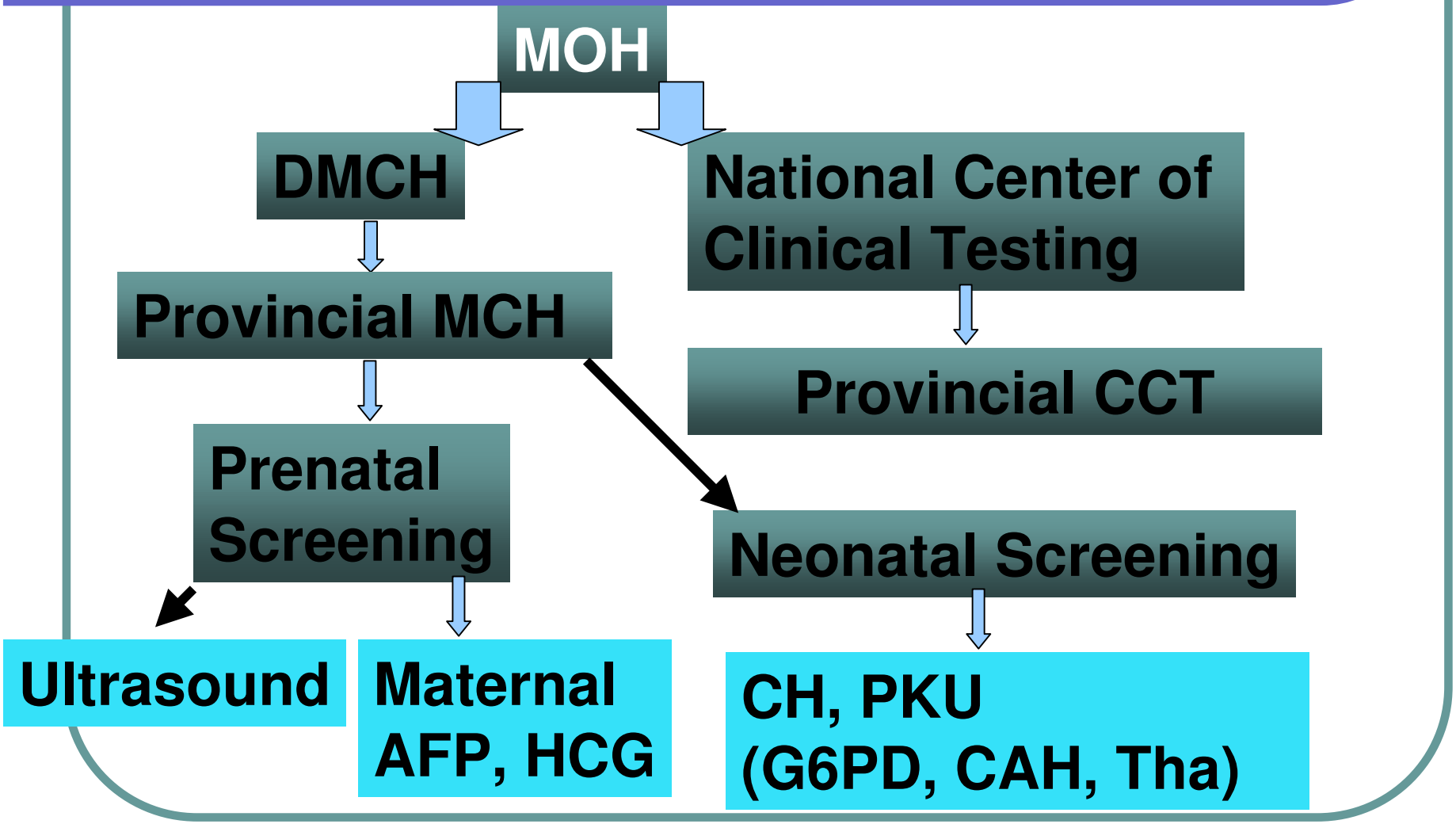
Policy and Regulation

《中华人民共和国母婴保健法》
National Law of Maternal and Child Healthcare

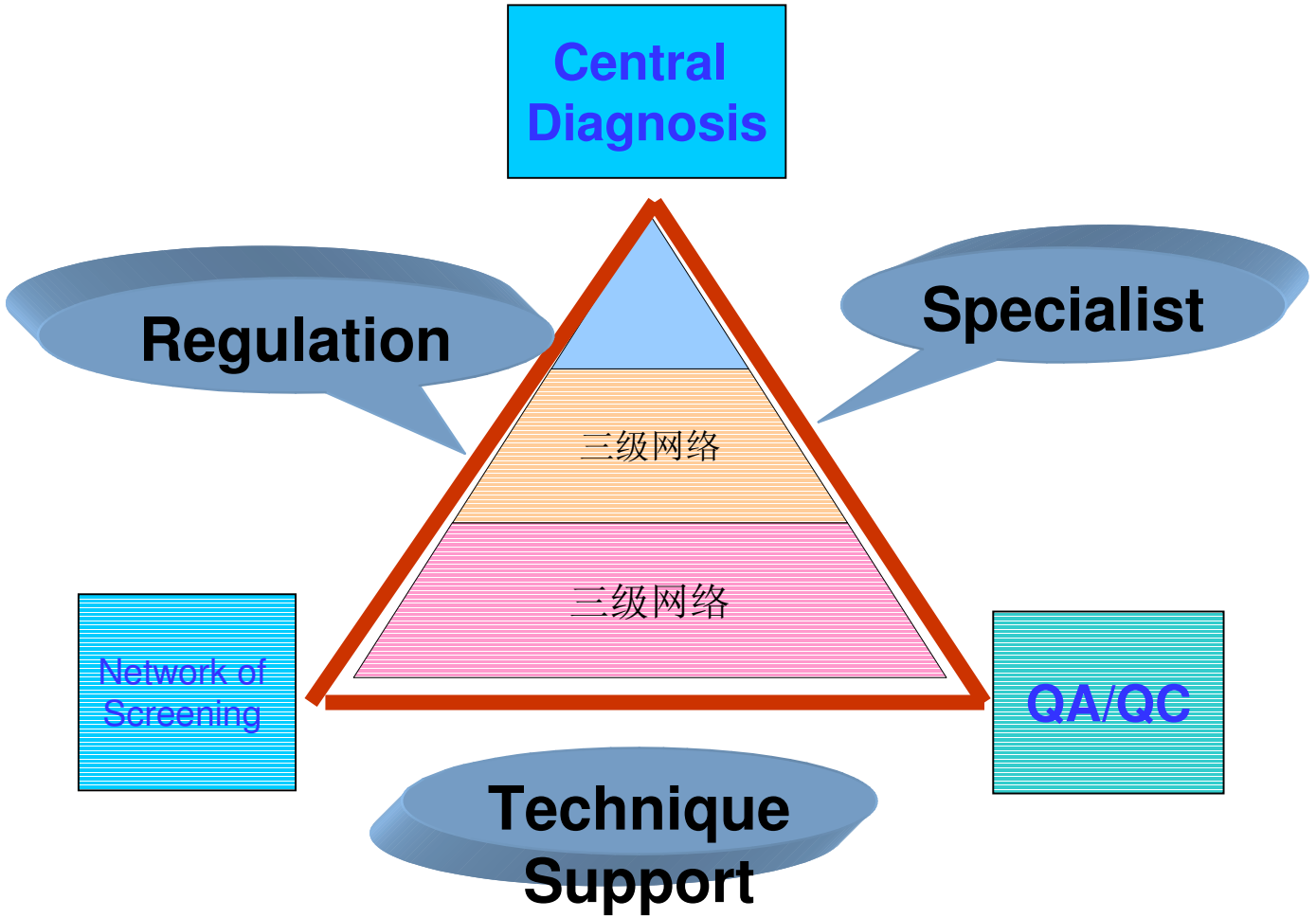
卫生部令《新生儿疾病筛查管理办法》
MOH Regulation: Neonatal Screening

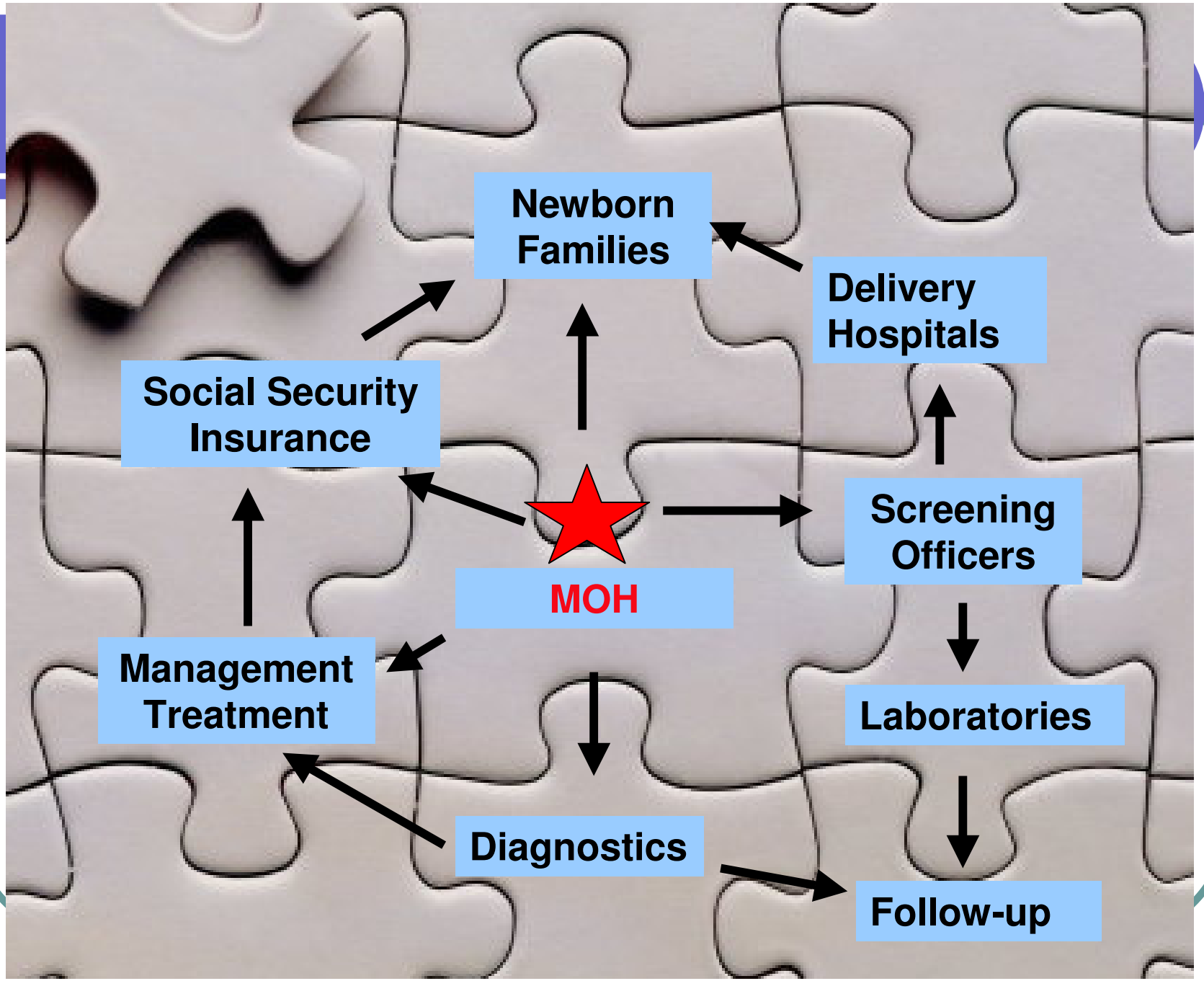
卫生部令《产前诊断管理办法》
MOH Regulation: Prenatal Diagnosis

National Mandatory Genetic testing

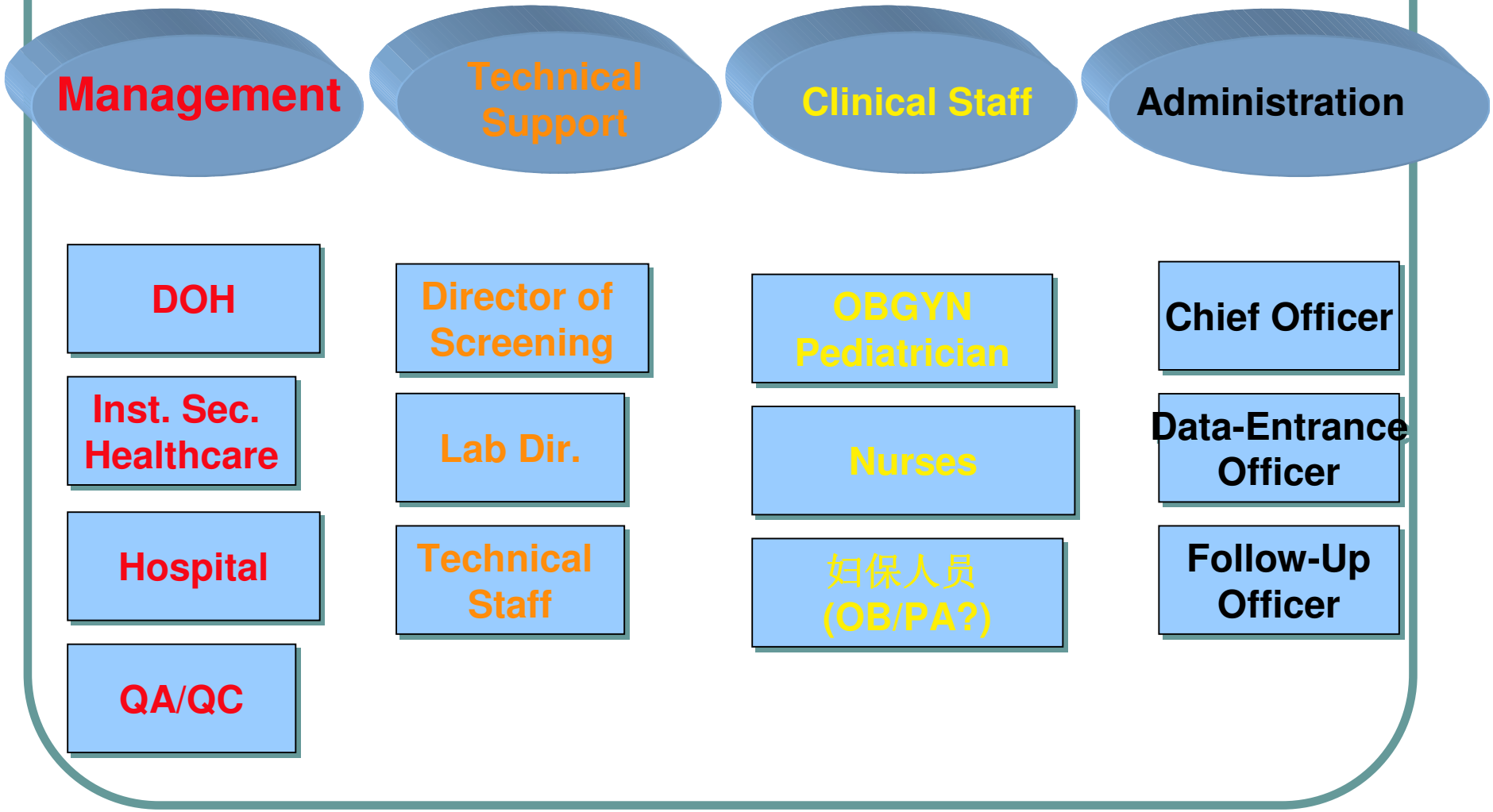


National “Triangle” Screening Model

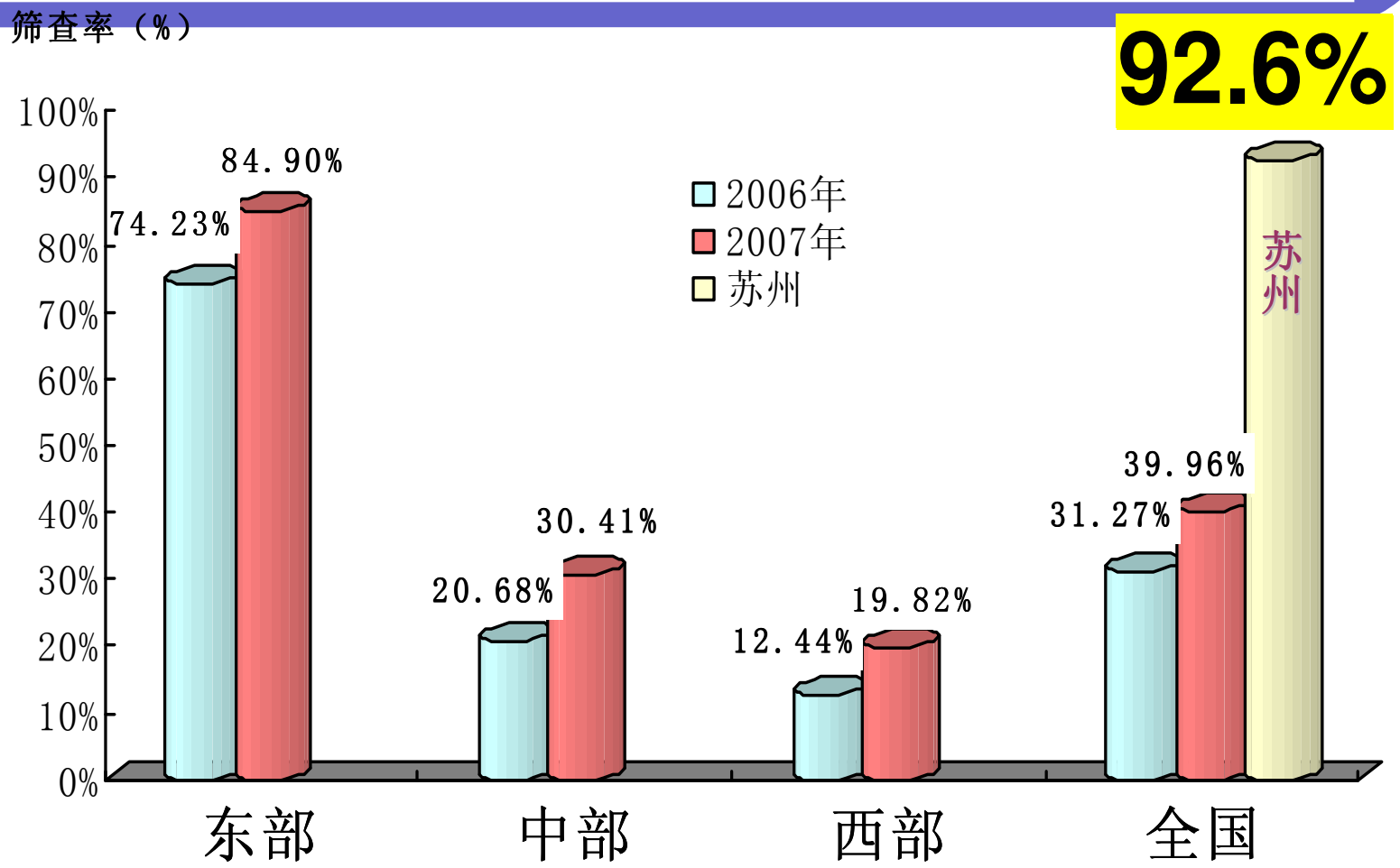



Newborn Families
Delivery Hospitals
Social Security Insurance
Screening Officers
MOH
Management Treatment
Laboratories
Diagnostics
Follow-up

“Clinical Geneticists?”

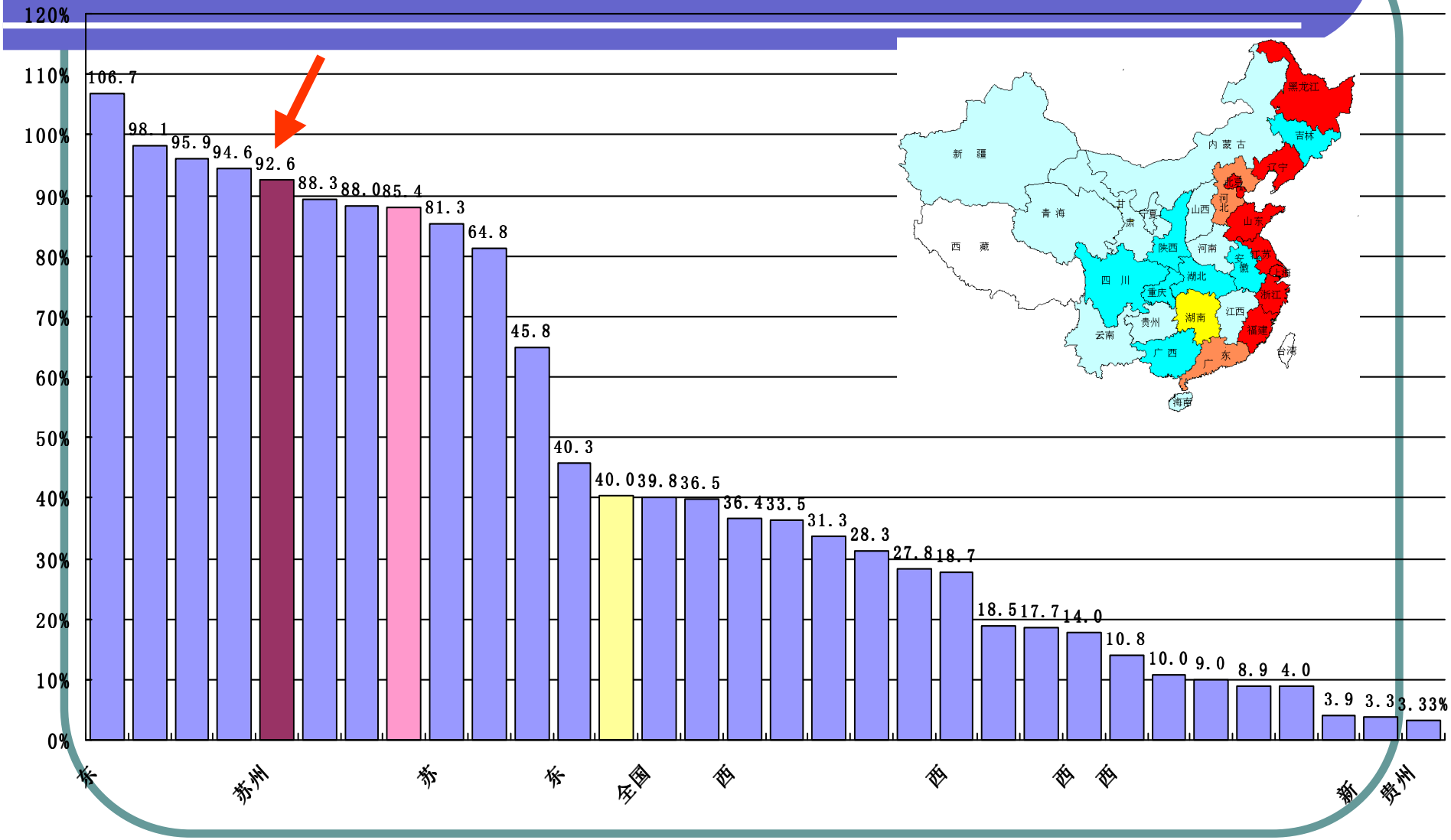


National Wide Newborn Screening (2006-2007)



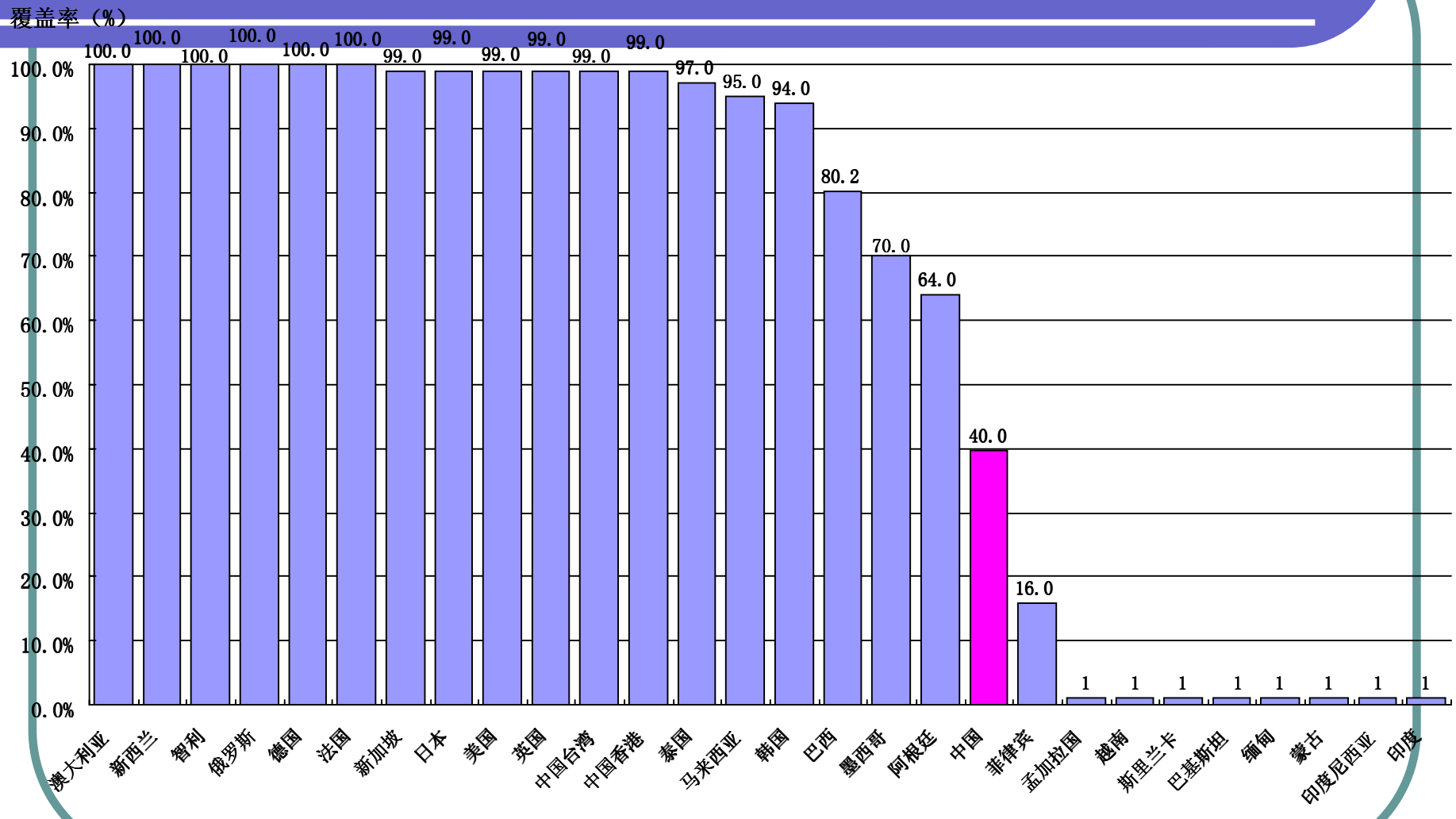
2006年和2007年全国东、中、西部地区筛查率情况

National Wide Newborn Screening (2007)



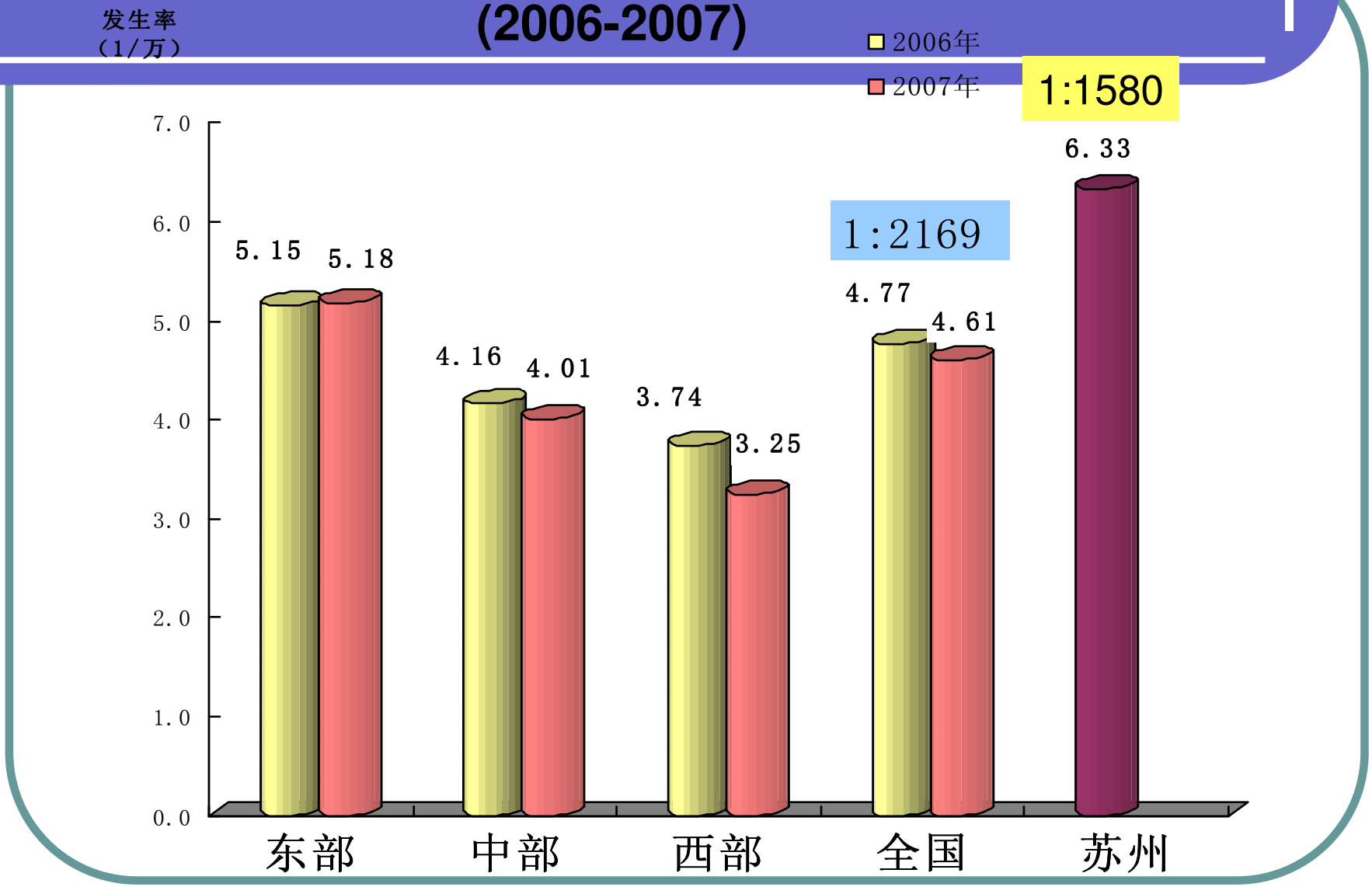
2007年全国　　新生儿筛查率情况(%)

World Wide Newborn Screening (2007)



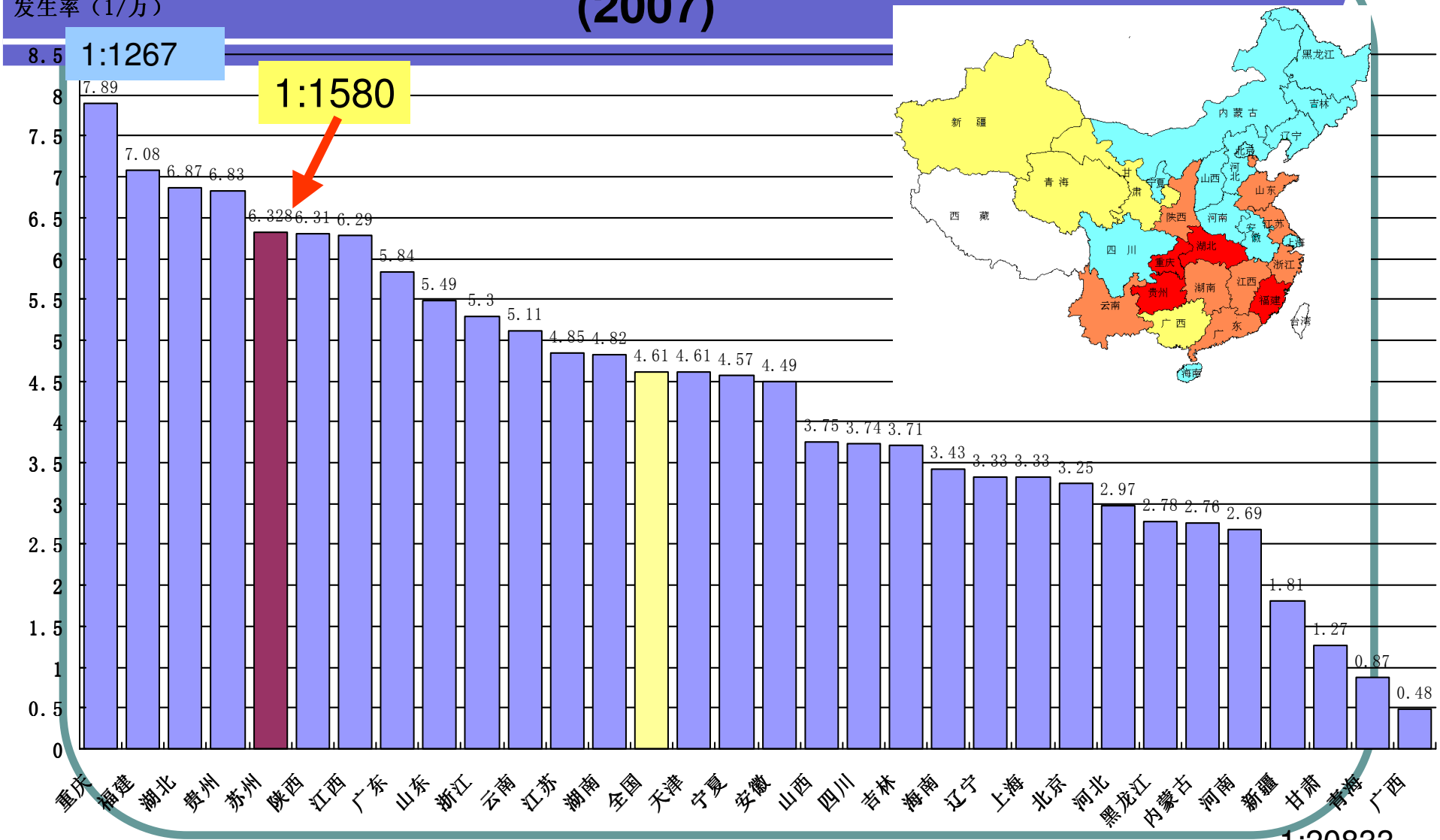
2007年世界主要国家新筛覆盖情况比较

Prevalence of National CH (2006-2007)



2006年和2007年全国各地区CH发生率情况

Prevalence of Provincial CH (2007)



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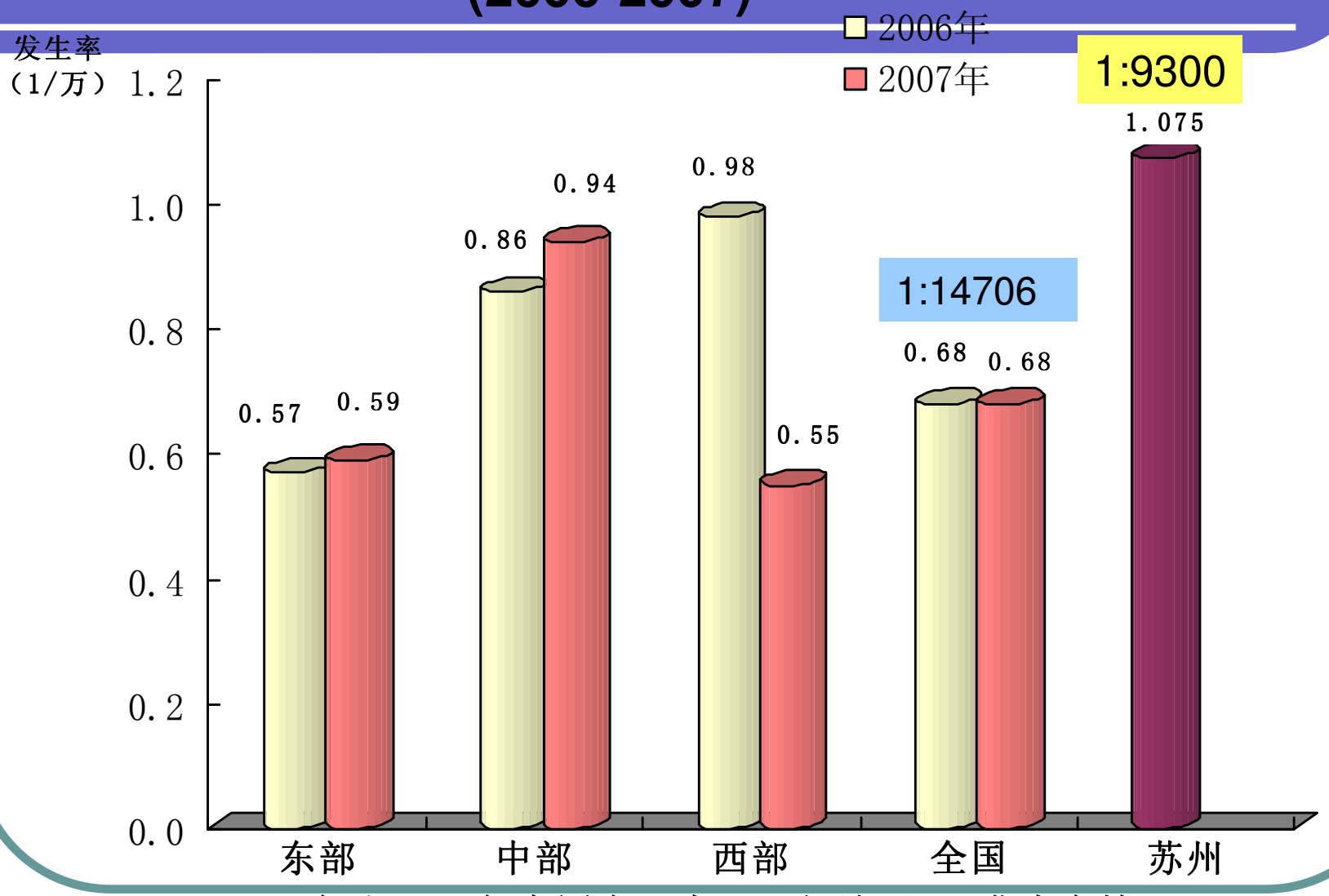
2007年全国各省新生儿甲低发生率情况

Prevalence of National PKU

(2006-2007)



2006年和2007年全国东、中、西部地区PKU发生率情况

Prevalence of Provincial PKU

(2007)

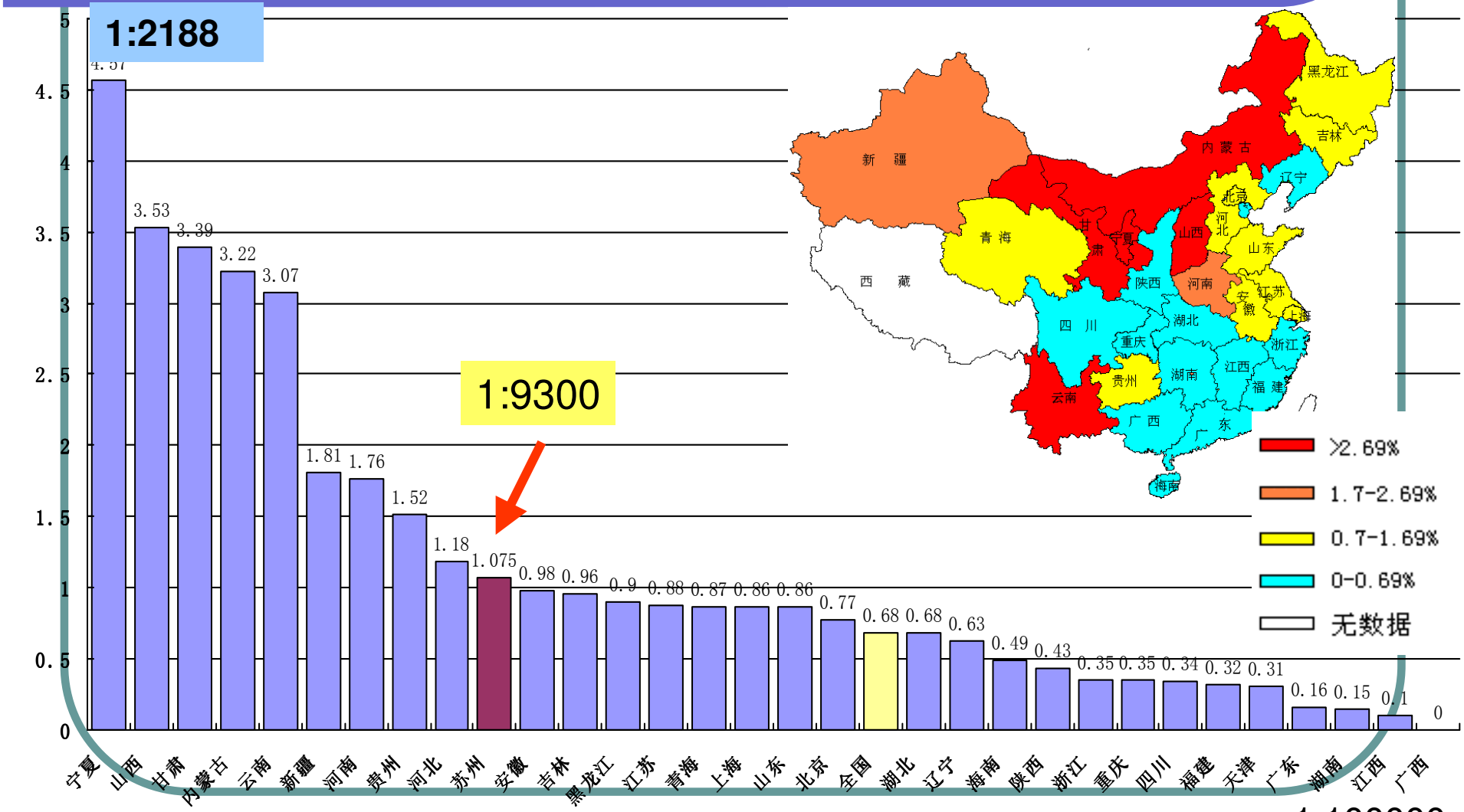


图37　2007年全国各省新生儿PKU发生率情况

Translational Medicine

The majority of genetic research conducted in hospitals or in medical schools

Genetic Testing of Common Diseases

分子细胞遗传

细胞遗传

分子诊断

新生儿筛查

实验基因治疗

神经遗传病

生化遗传

疾病检测诊断列表

相关站点链接

联系我们

1. 弗里德赖希共济失调(Friederich Ataxia 1, FA)
2. 齿状核红核苍白球路易斯核萎缩症(dentatorubral-pallidoluysian atrophy, DRPLA)
3. 强直性肌营养不良(myotonic dystrophy, DM)
4. 遗传性脊髓小脑性共济失调1型(spinocerebellar ataxia type 1, SCA-1)
5. 遗传性脊髓小脑性共济失调2型(spinocerebellar ataxia type 2, SCA-2)
6. 遗传性脊髓小脑性共济失调3型(spinocerebellar ataxia type3, SCA-3)
7. 遗传性脊髓小脑性共济失调6型(spinocerebellar ataxia type6, SCA-6)
8. 遗传性脊髓小脑性共济失调7型(spinocerebellar ataxia type7, SCA-7)
9. 遗传性脊髓小脑性共济失调8型(spinocerebellar ataxia type8, SCA-8)
10. 遗传性脊髓小脑性共济失调10型(spinocerebellar ataxia type10, SCA-10)
11. 遗传性脊髓小脑性共济失调12型(spinocerebellar ataxia type12, SCA-12)
12. 遗传性脊髓小脑性共济失调17型(spinocerebellar ataxia type17, SCA-17)
13. 亨廷顿(Huntington disease, HD)
14. 延髓脊髓性肌萎缩(SBMA)
15. 脆性X综合征(Fragile X syndrome)
16. Duchenne/Becker型肌营养不良(Duchenne muscular dystrophy, DMD)
17. 脊肌萎缩症(spinal muscular atrophy, SMA)
18. 腓骨肌萎缩症(Charcot Marie Tooth disease, CMT)
19. 镰刀形贫血(Sickle Cell Disease,SCD)
20. 凝血因子V缺乏(Factor V)
21. 肌张力障碍(Dystonia)
22. 先天性无虹膜症(Aniridia)
23. 核纤层蛋白综合征(Laminopathies)
24. 帕德-维利/安吉曼综合征(Prader-Willi/Angelman syndrome)
25. 迪乔治综合症(DIGeorge syndrome)
26. 21三体综合症(Down Syndrome)
27. α地中海贫血(α-thalasemia)
28. β地中海贫血(β-thalasemia)
29. 甲型血友病(Hemophilia A)
30. 乙型血友病(Hemophilia B)
31. 血管性假血友病(Von Willebrand Disease)
32. X-连锁智力低下(X-linked Mental Retardation)
33. 假肥大性肌营养不良症(Pseudohypertrophic Muscular Dystrophy)
34. 肢带型肌营养不良症(Limb-girdle Muscular Dystrophy)
35. 成人型多囊肾病(Adult Polycystic Kidney Disease, APKD/PKD1)
36. 婴儿性多囊肾病(Polycystic Kidney, Infantile)
37. Alport综合征(Alport Syndrome)
38. GJB2遗传性神经性耳聋(GJB2-related Genetic Sensorineural Deafness)
39. 苯丙酮尿症(Phenylketonuria, PKU)
40. 肝豆状核变性(Hepatolenticular Degeneration)
41. G6PD缺陷症(Glucose-6-phosphate Dehydrogenase Deficiency)
42. 先天性肾上腺皮质增生症(Congenital Adrenal hyperplasia)
43. 肾上腺脑白质营养不良(X-linked Adrenoleukodystrophy,ALD)
44. 甲基丙二酸血症(Methylamalonic Acidemia)
45. 神经元蜡样脂褐质储积症(Neuronal Ceroid Lipofuscinosis)
46. 遗传性脑白质病(Hereditary Leukoencephalopathy)
47. 线粒体脑肌病(Mitochondrial Encephalomyopathy,ME)
48. X连锁严重联合免疫缺陷(X-linked Severe Combined Immunodeficient)
49. Leber遗传性视神经病变(Leber Hereditary Optic Neuropathy, LHON)
50. 视网膜色素变性(Pigmentary Degeneration of the Retina, Retina Pigmentosia, RP)
51. 视网膜母细胞瘤(Retinoblstoma, RB)
52. 神经纤维瘤I型(Neurofibromatosis Type I)
53. 结节性硬化(Tuberous Sclerosis)
54. 成骨不全(Osteogenesis Imperfectas, OI)
55. 帕金森病(Parkinson Disease, PD)
56. 肌张力障碍(Dystonia)
57. 肌阵挛性肌张力障碍(Myoclonic Dystonia)
58. 先天性双侧输精管缺如(Congenital Bilateral Absence of Vas Deferens)
59. 遗传性精子生成障碍(Spermatogenic Failure)
60. 软骨发育不全(Achondroplasia)
61. Gaucher病(Gaucher Disease)
62. 糖原储积症(Glycogen Storage Disease)
63. 粘多糖病(Mucopolysaccharidosis, MPS)
64. 马凡氏综合征(Marfan Syndrome)
65. 家族性高胆固醇血症(Familial Hypercholesterolemia, FH)
66. 肥厚性心肌病(Hypertrophic Cardiomyopathies, HCM)
67. 孤独症(Autism)
68. 科-勒二氏综合征(Coffin-Lowry Syndrome, CLS)
69. 卡尔曼综合症(Kallmann Syndrome,KAL)
70. 染色体制备及G显带、C显带、N显带、SCE及微核制备
71. 多种疾病的淋巴细胞建库

各样品的采集要求:
分子诊断疾病:双亲外周血10ml，肝素抗凝。 其中5ml提DNA，5ml细胞培养;
先证者外周血10ml，肝素抗凝，其中5ml提DNA，5ml细胞培养。
细胞分子遗传检测：外周血5ml，肝素抗凝。
淋巴细胞建库：外周血6ml，肝素抗凝。
样品送北京大学医学遗传中心解剖楼230

Intended Survey: Geographically

North-Eastern

Northern (Headquarter)

Eastern

Southern

Mid-Western

South-Western

North-Western

Intended Survey: Personnel

- Program Director
  - Regional coordinators x4
    - Headquarter x1
    - North-Eastern/Western x1
    - Eastern/Southern x1
    - Mid-Western/Southern-Western x1
  - Provincial coordinators x ?

Intended Survey: Budget

- Personnel
- Travel
- Training
- Web-based database

Thanks for your attention !

- i) descriptive demographic and health care indicator data, epidemiological data including clinical and genetic epidemiology
- (ii) descriptive data of the national health care system, funding and infrastructure
- (iii) an inventory of genetic testing service settings and given service structures (e.g. integration of services into private or public health care-settings, services provided by research), funding of services, cost effectiveness, prioritisation and rationing, sustainability of service initiatives, structure, process and outcome data, quality issues, workload, professional training and education
- (iv) data on networking and interaction of services including already existing transnational activities
- (v) data on national/regional registries and biobanks
- (vi) description of legal and regulatory frameworks
- (vii) overview of research priorities in genetics/genomics funded by governmental resources
- (viii) role of patient organisations, public education and information
- (ix) bibliographic references