An Universal Platform for rare disease in China: from omics research to patient care

罕见病“平台“ 从组学研究到病人关怀

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China National GeneBank 国家基因库

Shenzhen, China, 8th Nov. 2014.

rare disease

- is any disease that affects a small percentage of the population.
- Most rare diseases are genetic **(80%)**, and thus are present throughout the person's entire life, even if symptoms do not immediately appear. Many rare diseases appear early in life, and about 30 percent of children **(30%)** with rare diseases will die before reaching their fifth birthday.[1] With a single diagnosed patient only, ribose-5-phosphate isomerase deficiency is considered the rarest genetic disease.
- 廣為人知的罕見疾病如苯酮尿症、重型地中海貧血、成骨不全症、黏多醣症、高血氨症、有機酸血症、威爾森氏症等
- 美国：患病率<1/1500（2002罕见病法案）；日本：患病率<1/2500
- 中国：患病率<1/50万；新生儿<1/10000。中国总患病人口1680万
- A disease may be considered rare in one part of the world, or in a particular group of people, but still be common in another.

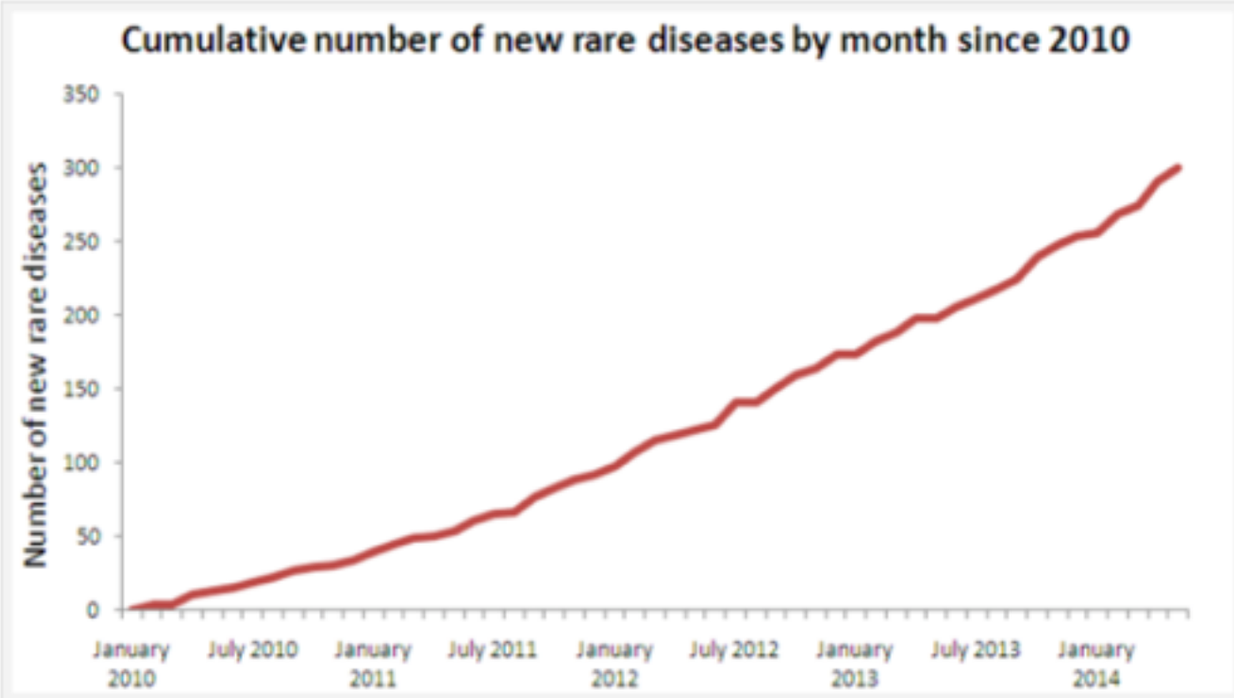
from wikipedia.org

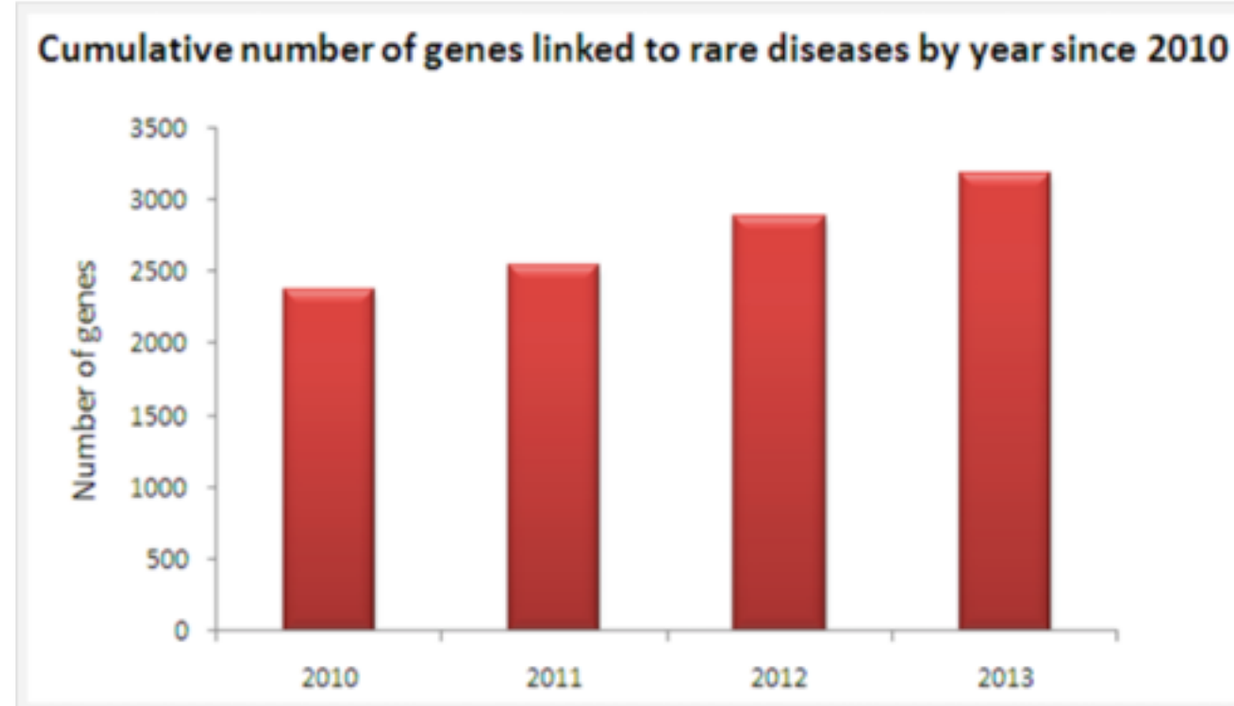
research in rare disease

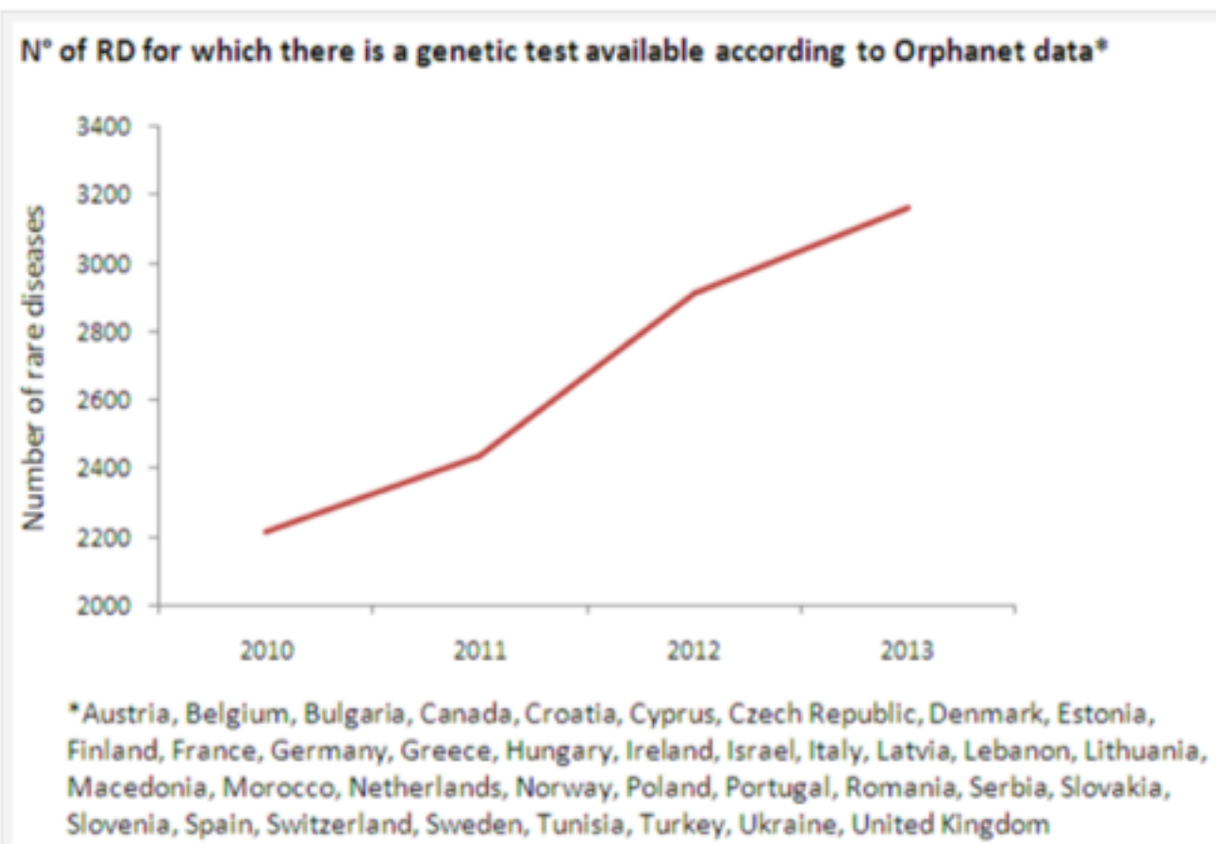
- ~6000-7000 (8000) different rare disease, but only less than 400 have relevant medical methods/ products

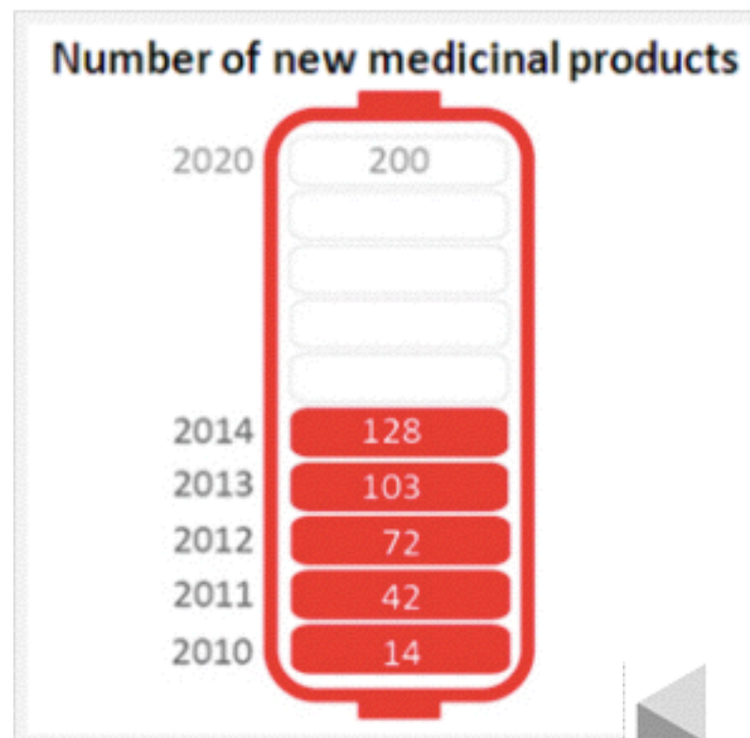
- 80% is genetic related. Therefore, a genetic screening might help to identify them in the different stages.

- 大部分的罕見疾病是經由突變或遺傳的基因缺陷而導致的先天性疾病，也有部分的罕見疾病至今尚未發現確實的致病原因。若父母雙方恰巧擁有相同的隱性致病基因、某一方家族有病史，或基因偶發地產生突變，出生的下一代即有可能出現基因異常的罕見疾病。









Almost 4,000 Projects Funded

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

hospital for rare disease

- 2011年2月，上海成立中国首个罕见病专科分会 – 上海市医学罕见病专科分会。
- Feb., 2011, Shanghai initialized the first rare disease specialist association (branch).
- 2011.12.22 上海选定交通大学医学院附属新华医院、复旦大学附属儿科医院、交通大学医学院附属瑞金医院为上海罕见病诊疗定点医院。与此同时，上海在罕见病医疗保障、疾病筛查、药物研发等方面都有新进展。
- Dec, 2011, Shanghai first time announced 3 hospitals will be official designated rare disease hospitals. The government starts to put a lots of attention into rare diseases.
- 2013年1月28日，华南首家罕见病门诊，深圳市儿童医院成立罕见病结节性硬化症门诊。
- Jan, 2013, the first rare disease out-patient was opened for tuberoussclerosis.

hospital for rare disease

- 在我国，罕见病误诊率达44%，确诊周期短则5年，长则30年——确诊后，往往也意味着死亡将至，75%的患者接受了不规范的治疗。鉴于目前全球仅1%的罕见病是可治的，上海确立的防治思路是先从可治的罕见病入手。

- in China, the wrong diagnostic rate for rare disease is 44%. The period for diagnostics may last from 5 years to 30 years. 75% of patients had wrong or not proper treatment.

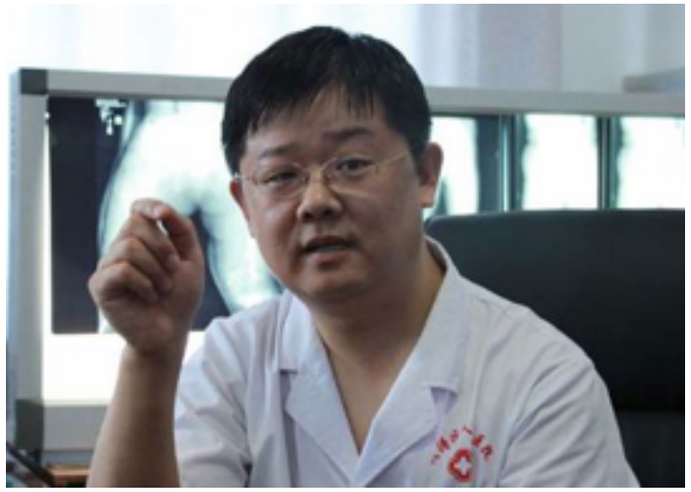
- 上海市医学会罕见病专科分会副主任委员马端指出：“罕见病其实是一种可防可治的疾病，它的预防为三级预防体制，一级针对没有怀孕之前的基因筛查，二级预防是怀孕以后到孩子生出来之前的筛查,三级预防是针对幼儿一岁之前的。越早查出来可以越早治疗，比如苯丙酮尿症，早期检查出来后患儿按照疾病特征服用可服用的食物，就可以维持终身的健康和正常。”

- rare disease, we should do some screening in different stagtes: genetic screening before pregnancy, prenatal diagnostic and early treatment after baby delivered (such as PKU).

Hospitals
good at rare disease

- 天津医院小儿骨科病房，任秀智

- Xiuzhi REN, OI, Tianjin Hospital
  pediatric department of orthopedics ward



中国罕见病
Rare Disease in China

public org in rare disease

美国罕见疾病组织由患有罕见病的个人和病患者的家庭在1983年组织成立。

The National Organization for Rare Disorders, established 1983. for research, education, collaboration.

1986年成立的遗传联盟列出了大约1200中罕见病的信息和支持团体。

全球基因计划是世界上最重要的罕见病与遗传病患者维权组织之一，这一非营利组织由R.A.R.E.（Rare disease, Advocacy, Research and Education）团队领导[23]。全球基因计划推动满足罕见病和遗传病群体的需要[24]。。2009一些罕见病患者双亲和基金会发起一次草根运动，催生了500余家全球性组织。全球基因计划提出“基因与牛仔裤”（"genes and jeans”，英语中两词谐音）的概念，广泛推动社会注意罕见病和遗传病团体的需求。这一组织已经发动了多项极具创意的罕见病和遗传病宣传运[25][26][27]。

加拿大罕见病组织是一个全国性组织，代表加拿大受到罕见病影响的人群，宗旨是为罕见病患者发出统一的声音，推动为罕见病患者建立社会在医疗系统与实行专门的卫生政策[28]。Canadian Organization for Rare Disorders (CORD).

希腊罕见病患者的组织希腊罕见病联盟（Greek Alliance of Rare Diseases）。

public org in rare disease

China:

瓷娃娃 CHINA-DOLLS

funded by Osteogenesis Imperfecta (OI) patients.

Our Motto : Love is still strong.

罕见病发展中心
Chinese Organization for Rare Disorders (CORD)

Love is not rare because of you.

more and more…

- Clinical laboratory standards for next-generation sequencing: Clinical exome and genome sequencing
- Data sharing in large research consortia
  - BIG DATA (because of OMICS)

OMICS research

- Whole genome resequencing
- Exome-capture sequencing
- Transcriptome
- Digital gene expression profiling
- MicroRNA
- Epigenome
- Pathogen
- MS-based Proteome
- MS/NMR-based Metabolism

DNA Level

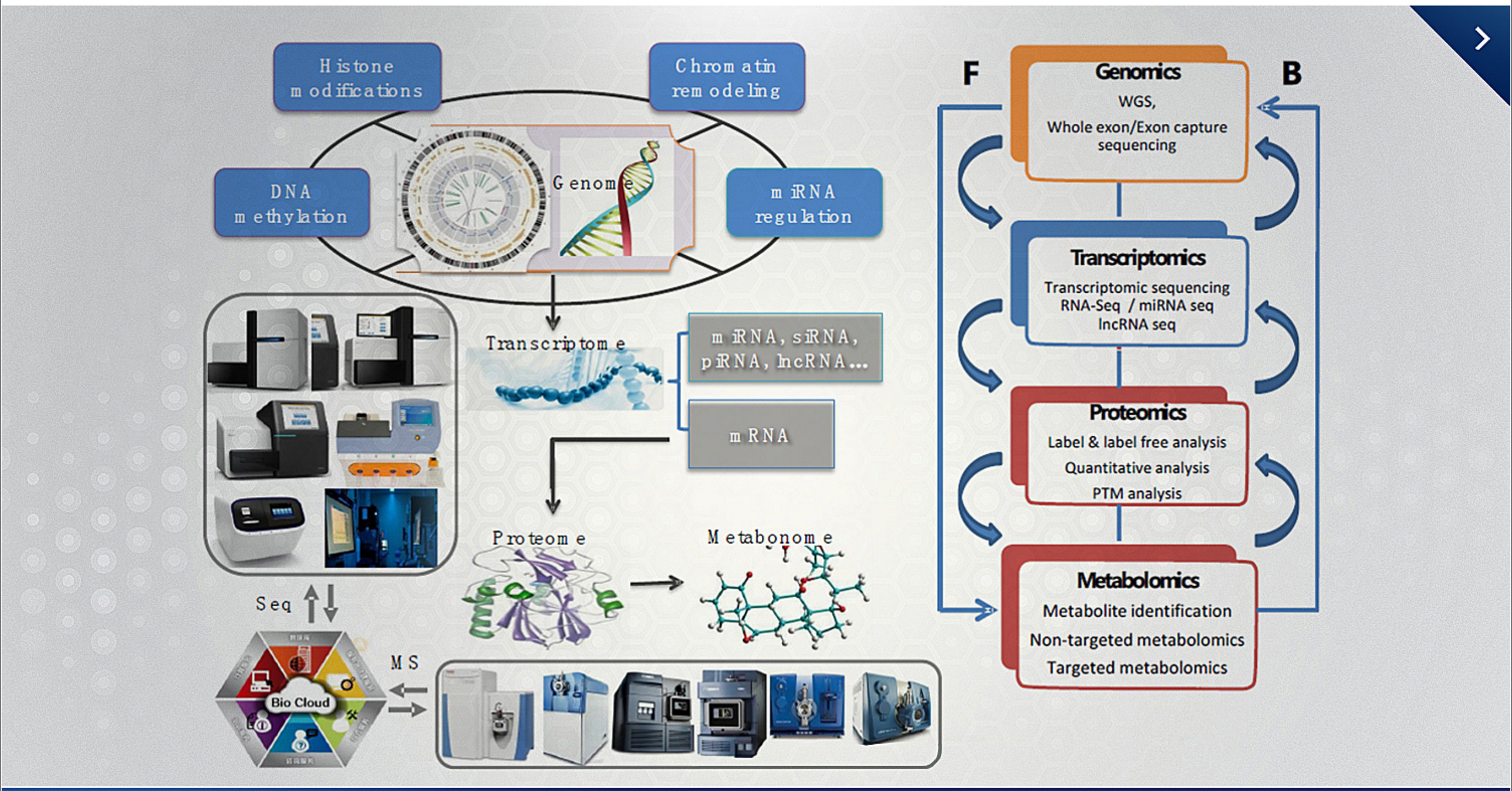
RNA Level

Epi

virus, etc

Protein Level

Metabolite Level



Systems Biology & BGI Trans-Omics Tools

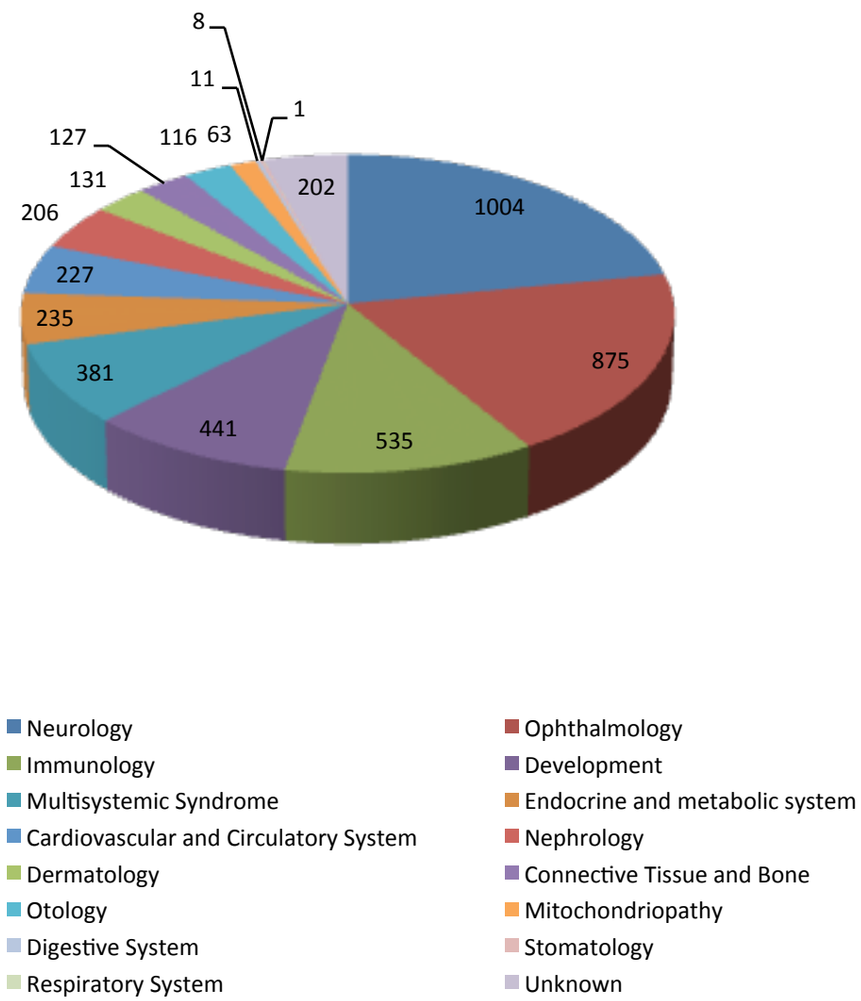
Progress of 1000 Mendelian Disorders Project

Until now, we have collaborated with about **150** organizations all over the world.

studies in BGI

We have collected about **6000** high quality samples covering more than **400** diseases.

About **4563** samples have been sequenced, and many are still under analysis and validation.



gender	case	control	unknown	total
Female	1045	248	206	1499
Male	1327	222	171	1720
unknown	232	42	1070	1344
Sequencing sample	2604	512	1447	4563

Disease	Gene	Inheritance Model	Sequenced Sample	Magazine	pubtime
spinocerebellar ataxias	TGM6	AD	5	Brain	2010/11/23
Acne Inversa (Hidradenitis Suppurativa)	NCSTN	AD	3	The Journal of Investigative Dermatology	2011/3/24
Familial Medullary Thyroid Carcinoma	RET	AD	6	Plos one	2011/5/31
High Myopia	ZNF644	AD	2	PLoS Genetics	2011/6/9
paroxysmal kinesigenic dyskinesias	PRRT2	AD	4	Brain	2011/11/26
Olmsted Syndrome	TRPV3	AD	3	American Journal of Human Genetics	2012/3/9
Retinitis pigmentosa	CYP4V2	AR	4	Plos One	2012/5/31
Leber congenital amaurosis	NMNAT1	AR	1	Nature Genetics	2012/7/29
mitochondrial DNA depletion sy ndrome-5	SUCLA2	AR	1	Molecular Genetics and Metabolism	2012/9/7
Punctate Palmoplantar Keratoderma	COL14A1	AD	3	J Med Genet	2012/9/13
Disseminated superficial actinic porokeratosis	MVK	AD	3	Nature Genetics	2012/9/16
Retinitis pigmentosa	SNRNP200	AD	1	Plos one	2012/9/19
Pure Hair and Nail Ectodermal Dysplasia	HOXC13	AR	4	American Journal of Human Genetics	2012/10/11
Polycystic Kidney Disease	PKD1	AD	2	Gene	2012/12/21
Osteopetmsis	CLCN7	AD	2	Gene	2013/1/4
Oculocutaneous albinism	SLC24A5	AR	3	The Journal of Investigative Dermatology	2013/1/30
Dyschromatosis universalis hereditaria	ABCB6	AD	4	The Journal of Investigative Dermatology	2013/3/21

We have jointly published 63 papers.
And we have still about 37 papers in preparation or submmition.
We have 7 authorized patents, and another 74 patents in the applying procedure .

Novel genes: ~60, published (24)
known gene with novel mutants: ~140, published (63)

Han’s

- challenge 2020,
- connecting databases
- matchmaking patients
- technologies
- “exome sequencing is much better diagnostic than me (for my 30 years experience).

aboard

- Standard
- Funding
- Scientists
- Network
- Drug development
- More friendly

in China

- Doctors with tremendous experience but lacking communication with aboard.
- Big patient population size
- Many good non-profit organizations

罕见病非盈利组织（病友会） Rare Diseases - Non-profit Organization

中国罕见病
Rare Disease in China

站内搜索

hanjianbing.org Go

罕见病百科 Database | 资讯中心 News | 罕见病政策 Policies | 罕见病组织 Organizations | 资源中心 Resources | 关于我们 About Us

罕见病患者信息登记

欢迎罕见病患者和家属参与登记，以便更好获取信息支持和服务，并为罕见病医疗和社会保障政策尽快出台作出贡献

医疗地图

通过便捷的检索工具，按疾病名称或地区查找中国境内提供罕见病诊断、检测和治疗的相关机构及联系方式

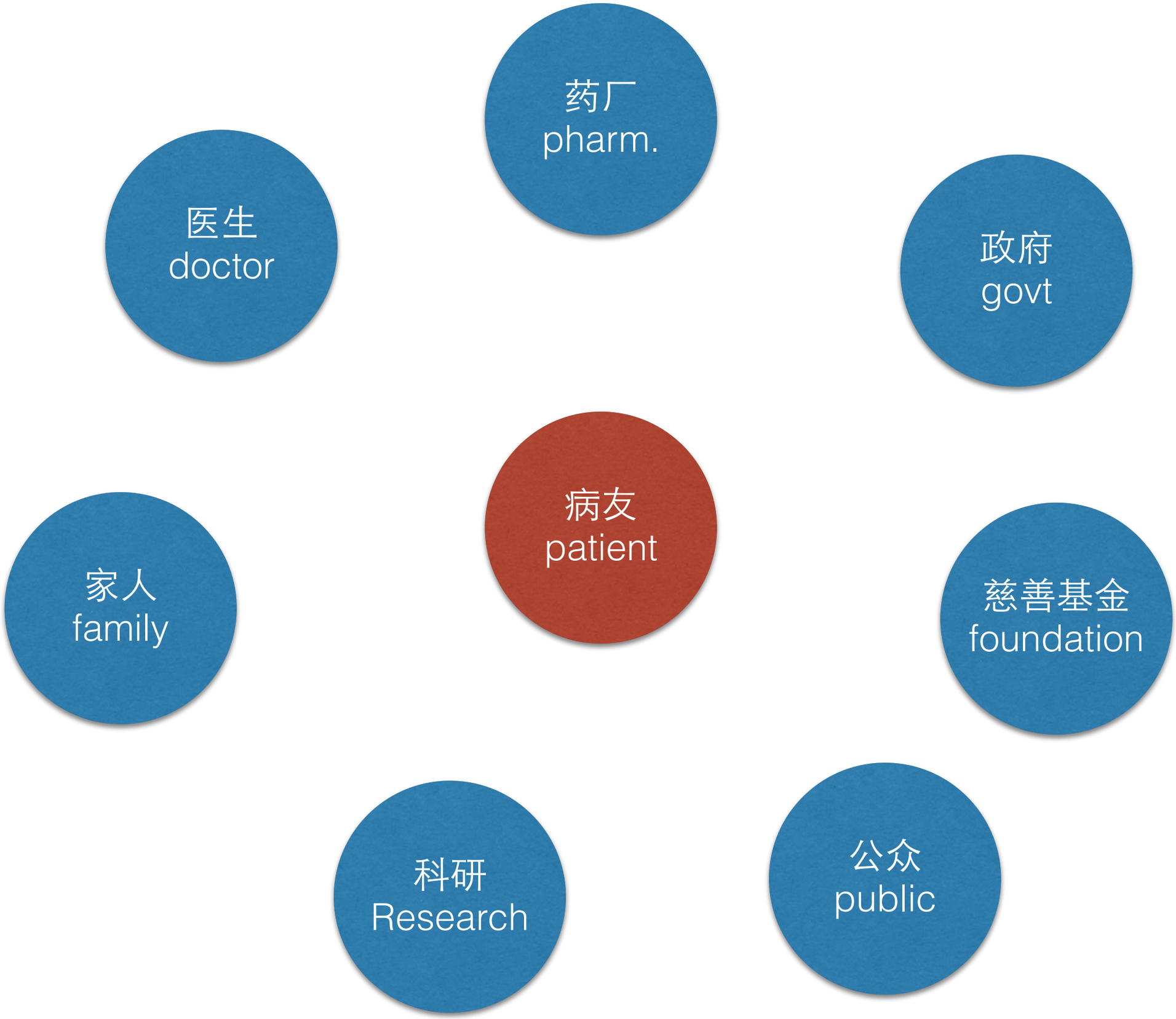
国际罕见病日

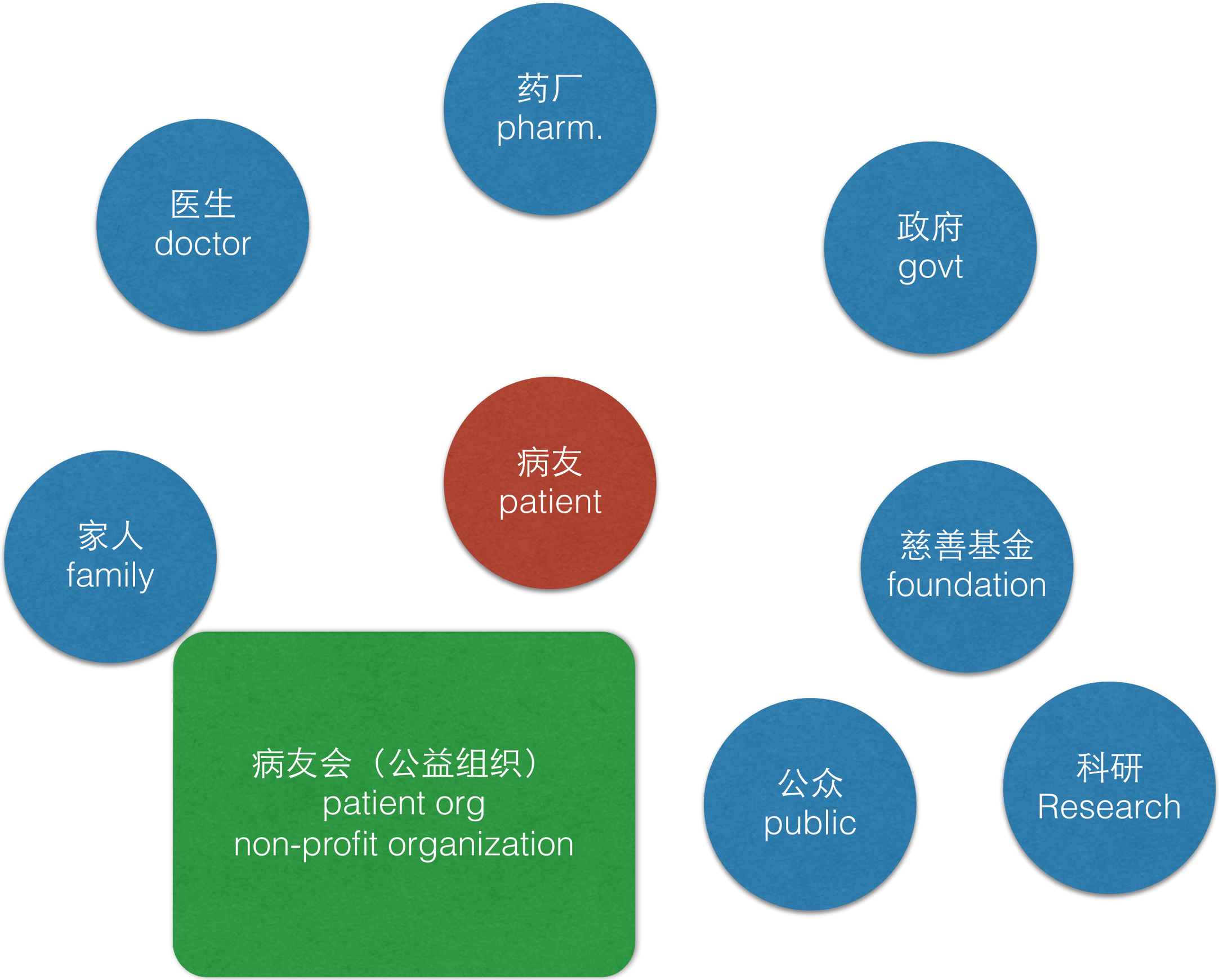
2月29日（或2月的最后一天）是国际罕见病日，旨在促进社会公众和政府对罕见病问题的关注

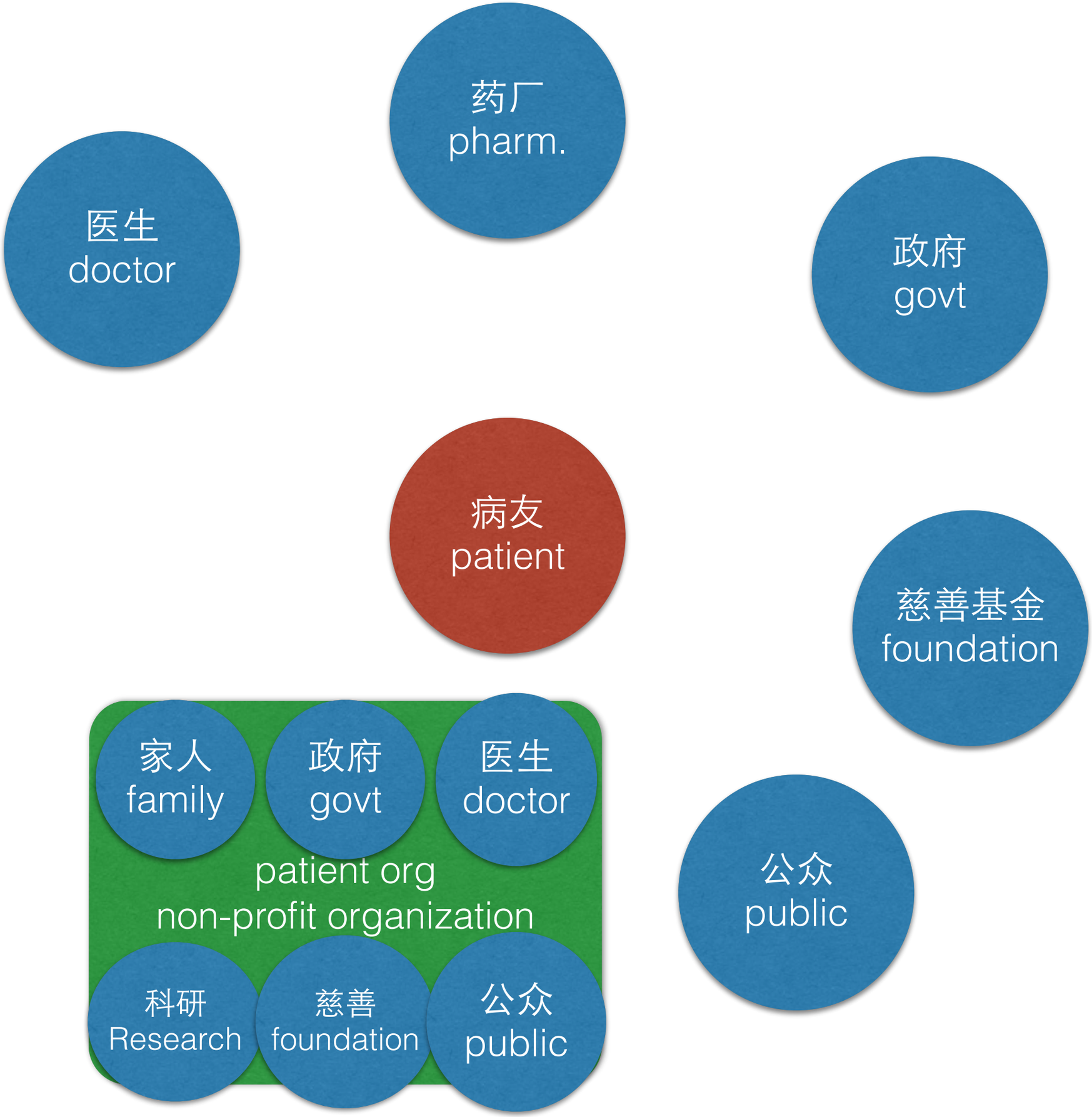
罕见病“中心” Chinese Organization for Rare Disorders (CORD)

patient care

关爱病人


药厂
pharm.
医生
doctor
政府
govt
病友
patient
家人
family
慈善基金
foundation
科研
Research
公众
public


药厂
pharm.
医生
doctor
政府
govt
病友
patient
家人
family
慈善基金
foundation
病友会（公益组织）
patient org
non-profit organization
公众
public
科研
Research


药厂
pharm.
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政府
govt
医生
doctor
patient org
non-profit organization
科研
Research
慈善
foundation
公众
public
公众
public

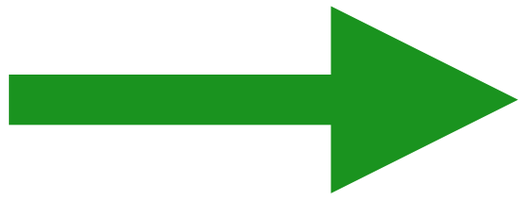
做点什么?

do something?

- 做科研，找病因，提供检测产品
  - Scientific Research Projects, Find causative mutant-genes and providing diagnostic products/service.
- 给钱 Donation.
- 关注关爱 Love and Care
- 更多的人了解罕见病。encouraging more people to know rare disease

Patient Care

- 公众支持和教育
  public support and education
- 政策法规
  policy and regulation
- 病友家庭
  patient family support
- 疾病研究
  research in rare disease
- 医院关怀／特殊医院
  special hospitals



正确的诊断
Correct diagnostic

治好／控制病
Right Treatment (no cure?)

不生有病的孩子
help on baby

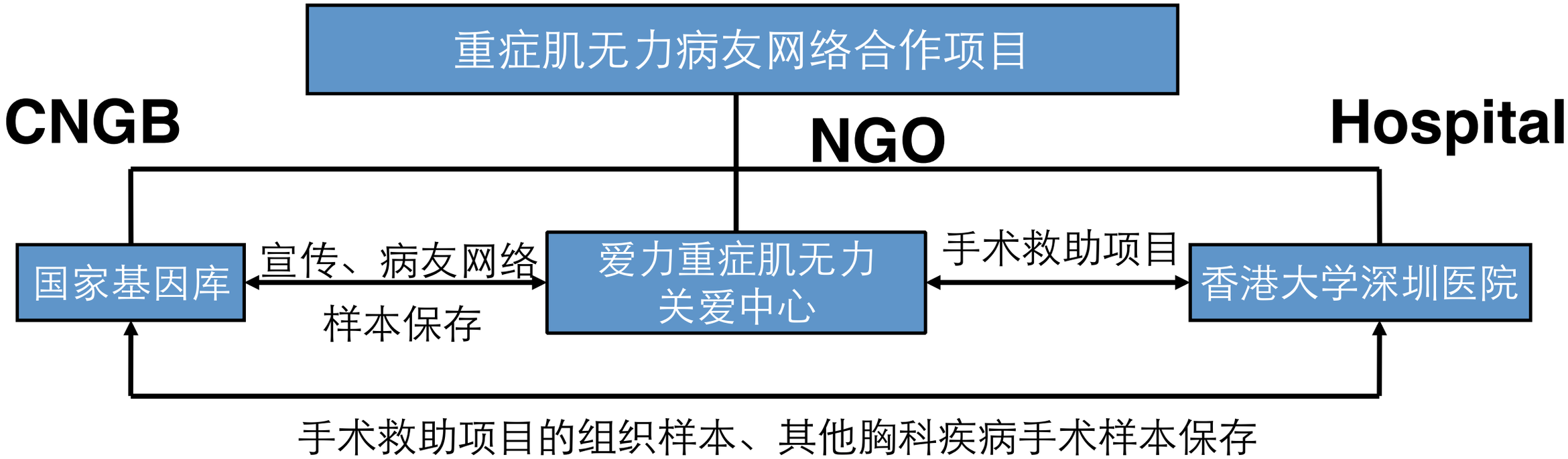
生活质量提升
improve life quality

An Universal Platform for rare disease in China:
from omics research to patient care

罕见病“平台“
从组学研究到病人关怀

重症肌无力病友网络项目规划与进展

国家基因库 CHINA NATIONAL GENEBANK



规划与进展

- **宣传**：赞助爱力中心6月病友大会宣传物料及配合宣传（已完成）
- **病友网络**：搭建爱力重症肌无力关爱中心门户网站及多维数据库（14年7月-15
- **样本保存**：港大深圳医院样本保存（已开展合作，计划2014年保存50-80例胸科疾病手术组织样本，其中10-20例为救助项目样本）

初级版本 **version 1.0**

logo+口号+400电话

注册/登陆

了解疾病　资讯中心　就医助手　与疾病相处　志愿者之家　关于我们

蓝色纸飞机

“蓝色纸飞机（coffeebar）”定位为：以商业模式运营校园公益，创业，跨界事业。主要...

—— 作者：aplane

关于疾病

欢迎罕见病患者和家属参与登记，以便更好获取信息支持和服务，并为罕见病医疗和社会保障政策尽快出台作出贡献

就诊地图

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患者注册

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最新资讯

FD House：Roger Vivier创意总监的家

设计　2014-01-20　4

runo Frisoni是鞋履设计大师，也是法国传奇鞋履品牌Roger Vivier的现任创意总监，他在鞋履设计中展

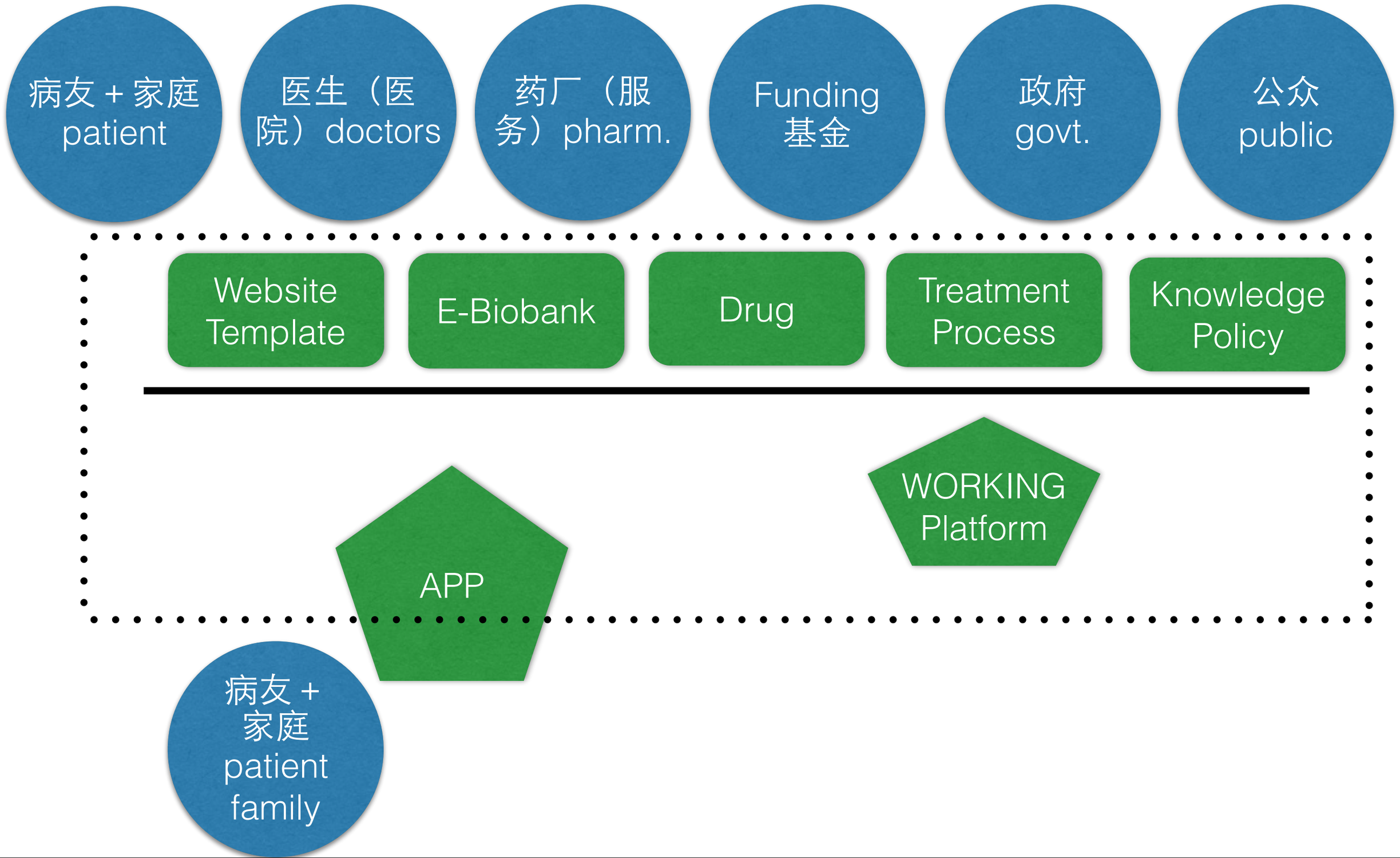
医生专栏　资料下载

1. 无蓝海蓝美不胜收 ——希腊Katikies酒
2. Carl Jurisch's 1957 Motoplan Con
3. Louise Campbell家居設計 Louise
4. 2013 TAIWAN DESIGN EXPO x
5. 非辣调味瓶：Ajori Cruet
6. Bri.Buckley剪貼畫作品 Arts by Bri.Bu
7. 全球知名的香港玩具公司threeA （3A）
8. Happy Pills 创意糖果店品牌形象设计 |
9. 韩国首尔江南区GA ON JAI 住宅
10. Stonehenge Visitor Centre by

爱力　支持我们

- 模版 – template website for every organisation
  - 支撑罕见病公益组织、病友会 to support all NGO
- 一个系统 - a system
  - 连接各病友会公益会之间关系。to connect NGOs
  - 连接病友之间关系 to connect patients
  - 连接病友和药厂、医疗服务之间关系 to connect with Pharm.
  - 连接病友和医生／医院之间关系 to connect with doctors
- 一个应用 - an application
  - 做到控制病、不生有病的孩子、……control diseases, prevention

an universal platform for all



3 NGOs
5,000 patients
3 hospitals
2 pharm.
20 research institute
2 governments

A report for rare disease:

status aboard
tax deduction
new clinical trial regulation rule
govt. protection plan

更多的了解罕见病
帮助他／她们

Let’s help them
from know more about rare disease