



SHANGHAI FIRST  
MATERNITY AND INFANT HOSPITAL  
— 1947 —  
上海市第一妇婴保健院

# Prenatal diagnosis of rare genetic diseases: what doctors want from government and industry

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# Rare genetic disease is not a disease

[Http://www.51mch.com](http://www.51mch.com)

- Rare genetic diseases are medical conditions, BUT, they are more like social problems and challenges



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# An email from a desperate mom

[Http://www.51mch.com](http://www.51mch.com)

- 段教授，您好！

我是两个孩子的妈妈，但不幸的是，我的两个孩子都患了同一种罕见病——窒息性胸廓发育不良(thoracic insufficiency syndrome, TIS)。胸廓的严重发育不良已经压迫到孩子的肺部、心脏和肝脏。我带着他们四处求医，但是医生都没有好的办法进行手术。我在网上查到您要去参加深圳第二届罕见病研究联盟会议，希望您能帮我一起寻找好的治疗的方式。我把孩子的一些照片和CT以及诊断书发给您，我们现在住在广州，如果需要任何补充的材料，请您随时与我们联系。

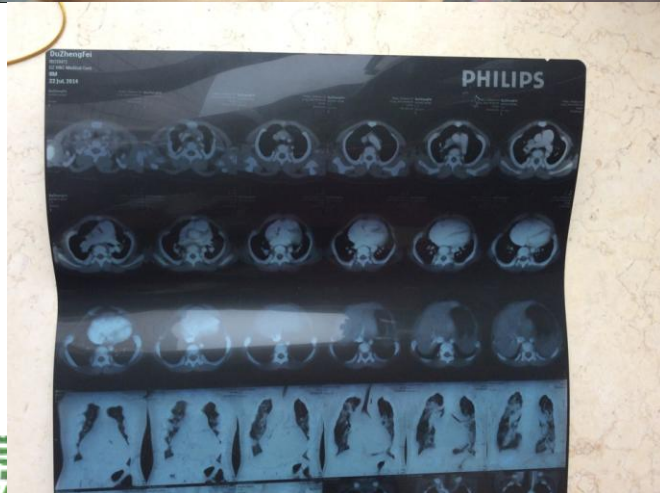
孩子焦虑的妈妈



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# What we want from government

[Http://www.51mch.com](http://www.51mch.com)

- The government should build a platform for the development of techniques and products related to the diagnosis and management of rare genetic diseases.
- The government should sponsor rare genetic diseases related basic and translational researches.



# What we want from government

[Http://www.51mch.com](http://www.51mch.com)

- The government should pool the resources to build national and regional referral centers for rare genetic diseases and sponsor these centers.



# What we want from the industry

[Http://www.17mch.com](http://www.17mch.com)

- The industry should make synergetic efforts to develop solutions for rare genetic diseases, because it might take more than one platform and more than one company to diagnose and manage one rare genetic disease.
- Free transfer of patents to the developing countries in the field of rare genetic diseases should be encouraged.

# What we want from clinical genetics

[Http://www.51mch.com](http://www.51mch.com)

- The protocols for rare genetic diseases should be simple, less expensive.



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Thanks! 谢谢聆听!



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