新闻稿
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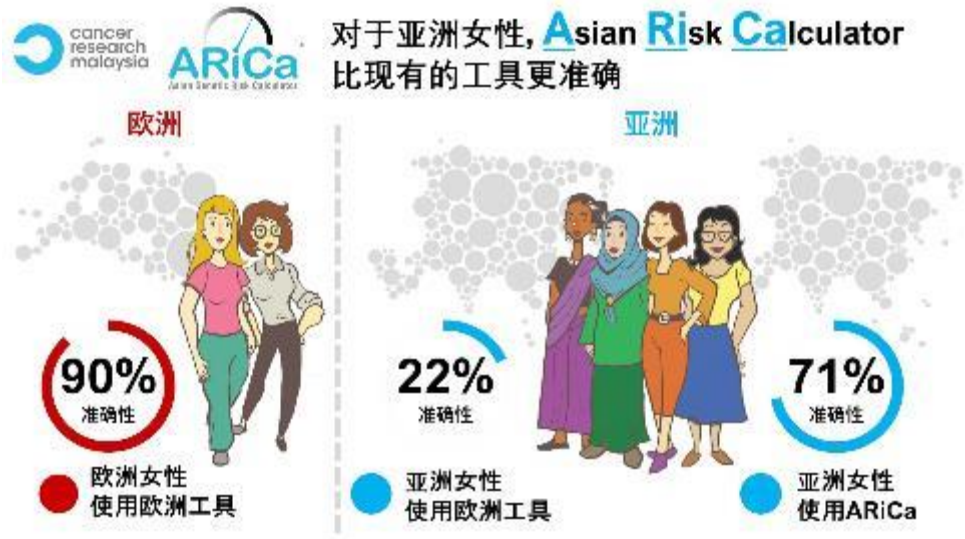
马来西亚团队研制能计算亚洲乳腺癌患者携带具有遗传性 *BRCA* 基因突变机率的工具

为亚洲女性所研制的 ARiCa 工具可以更准**确地**识别
BRCA 基因突变携带者和癌症患者的治疗方案，同时帮助提高后代的癌症预防。



(吉隆玻 3 日讯) 符合今年国际妇女节主题“打破偏见”的同时，马来西亚癌症研究基金会 (Cancer Research Malaysia) 通过研制亚裔遗传风险计算器 (ARiCa) 来协助妇女为自己的健康做出明智的决定。此工具能让患有乳腺癌的患者确认自己携带 *BRCA1* 或 *BRCA2* 基因（一**种**肿瘤抑制基因，有助于防止乳房、卵**巢和其他**类型的细胞过快或不受控制地生长和分裂） 突变的机率。

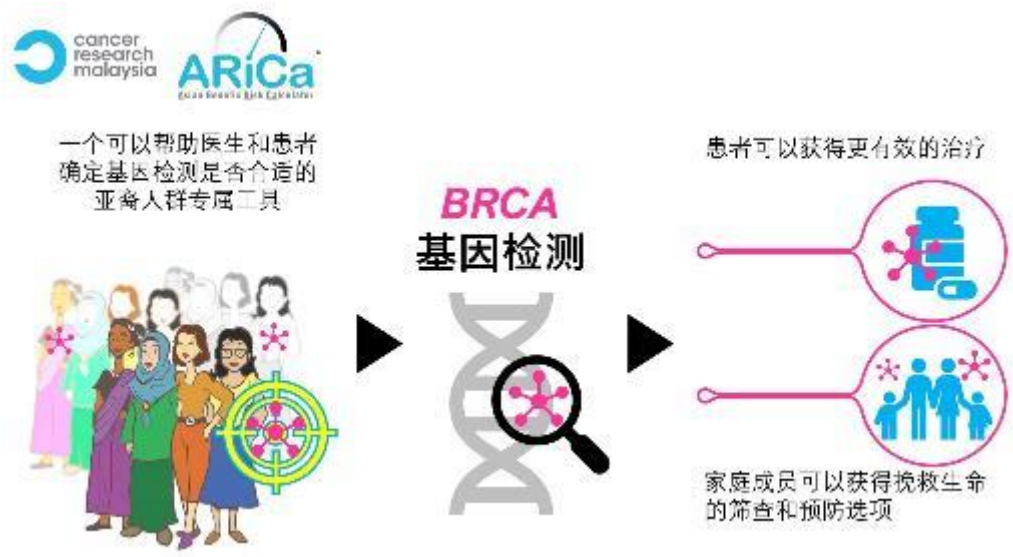
数据显示每二十五位乳腺癌患者当中，有一位将是 *BRCA* *突变*基因携带者。在这项研究之前，医生仅向年轻患者或有近亲患上乳腺癌或卵巢癌亲属的人推荐基因检测。但由于仅凭年龄和家族史并不能准**确**预测携带有缺陷基因的可能性，因此许多基因携带者没有进行基因检测，从而错过了对患者进行挽救生命的治疗和对近亲进行预防策略的机会。

为了克服这挑战，市面上推出了许多预测基因突变的预算器，但其中多数都是为欧洲女性所研制的，而对于亚洲女性来说往往不太准确。因此，亚洲病患与其亲属错失了了解自身基因遗传风险的机会，从而失去了癌症预防和准确治疗选择的可能性。

为了应对这一个挑战，马来西亚癌症研究基金会与剑桥大学、马来西亚大学、梳邦再也医疗中心、诺丁汉大学、新加坡基因研究所、新加坡国立大学、竹脚妇幼医院、陈笃生医院、新加坡国立癌症中心、新加坡中央医院以及樟宜综合医院合作分析了多于八千位来自马来西亚和新加

坡的乳腺癌患者的 BRCA1 和 BRCA2 基因。研究结果随后被用于研制一个工具以提供女性一个成为 BRCA 基因突变携带者的个人机率。

领导这项研究的首席研究员,马来西亚癌症研究基金会首席科学长拿汀巴杜卡张素芳教授说:“使用年龄和乳腺癌家族病史等标准，我们估计每年具有大约四千位乳腺癌新患者需要基因咨询和检测。但他们通常不考虑进行基因检测，因为大多数人认为他们不太可能遗传了有缺陷的基因。通过 ARiCa, 我们现在可以为每位女性提供成为 BRCA 携带者的个人机率，以便她们能够为自己的健康做出明智的选择，也希望医务人员们能够给患者提供更准确的医疗方案。”



这项研究联合首席研究员, 新加坡基因研究所的 Li Jingmei 博士表示：“这项在马来西亚与新加坡医院之间进行的合作研究让我们能够从华、巫、印裔乳腺癌患者的多种族人群中收集测试数据。这确保所研制的工具在亚洲族群中有相等的表现程度。”

剑桥大学癌症遗传流行病学中心主任和此项研究的共同领导研究员，道格拉斯.伊斯顿教授(Professor Douglas Easton)解释说：“直到最近，超过百分之九十的乳腺癌基因研究是在欧洲女性中进行的。在此研究中，我们成功研制一个可以为亚洲女性提供 BRCA1 或 BRCA2 基因突变携带者的个人机率的工具。我们的研究强调了了在各种族群中进行研究的重要性，以确保精准医学的发现能够公平地惠及所有人群。我们很高兴能够通过与马来西亚癌症研究基金会的合作以及惠康基金会和欧盟委员会的资金协助下将这项研究扩大到亚洲人群。”

剑桥大学的安东尼斯.安东尼奥教授，也是 BOADICEA 与 CanRisk（基于欧洲女性**开**发的工具）的首席**开**发人员补充说：“CanRisk 等工具现已在欧洲、北美、澳大利亚和其他国家广泛使用。这项研究为我们如何调整 CanRisk 以对亚洲国家的女性进行更准确的风险评估指明了方向。这些信息对于为低收入和中等收入国家调整这些工具可能特别重要，因为这些国家的资金和基础设施可能无法支持对每一位妇女进行筛查。”

梳邦再也医疗中心乳房外科顾问医生叶静娴教授解释说：“从医学角度来看，知道一位患者携带着 *BRCA* 基因突变可能对她们的手术管理有所改变。比方说，一位携带着 *BRCA* 基因突变的

患者有百分之五十的机率患上对侧乳腺癌。其一手术策略是切除双侧乳房并可选择立即进行乳房再造手术。由于 *BRCA* 基因携带者患上卵巢癌的风险也高达百分之三十至百分之四十，卵巢割除手术也可以同时进行。除此之外，医务人员也可以给患者提供一种针对 *BRCA* 基因突变，也被称为 PARP 抑制剂的新药，以提高患有早期或晚期乳腺癌携带者的生存率。由于我们无法为所有患者提供癌症遗传学服务，ARiCa 能**帮助我**们确认需要被转介进行基因咨询和检测的癌患者。"

马来西亚大学癌症研究所所长努尔艾莎教授(Professor Dr Nur Aishah Mohd Taib)强调说："虽然政府资助和公立医院无法常规提供预防性手术和靶向治疗，但确认 *BRCA* 携带者能帮助患者与其亲属了解自身的风险，以便他们及早发现癌症和规划其他的风险管理策略。我们在马来西亚大学已设立风险管理诊所超过十年了，以帮助有患病或无患病的携带者获得降低风险的医疗策略。我们希望 ARiCa 等工具能够让其它医院设立相类似的诊所。"

新加坡国立大学医院高级顾问迈克尔.哈特曼副教授(Professor Mikael Hartman)说："目前，由于亚洲女性癌症诊断的中位年龄较低，我们根据患者的诊断年龄与癌症家族病史等标准转介病患进行基因咨询与检测。即使在新加坡等亚洲发达国家，这也加剧了获得基因检测的挑战。因此，像 ARiCa 这样针对特定人群更准确地识别突变携带者的工具将帮助满足对癌症遗传学服务日益增长的需求。"

该研究已刊登在著名的医学杂志 Journal of Clinical Oncology 上。其发现将帮助医生和病患更有效地了解携带着 *BRCA1* 或 *BRCA2* 基因突变的机率。 对于安吉丽娜.朱莉等 *BRCA* 携带者来说，知道自己的终身风险可能意味着在咨询专家后做出不同的预防决定。

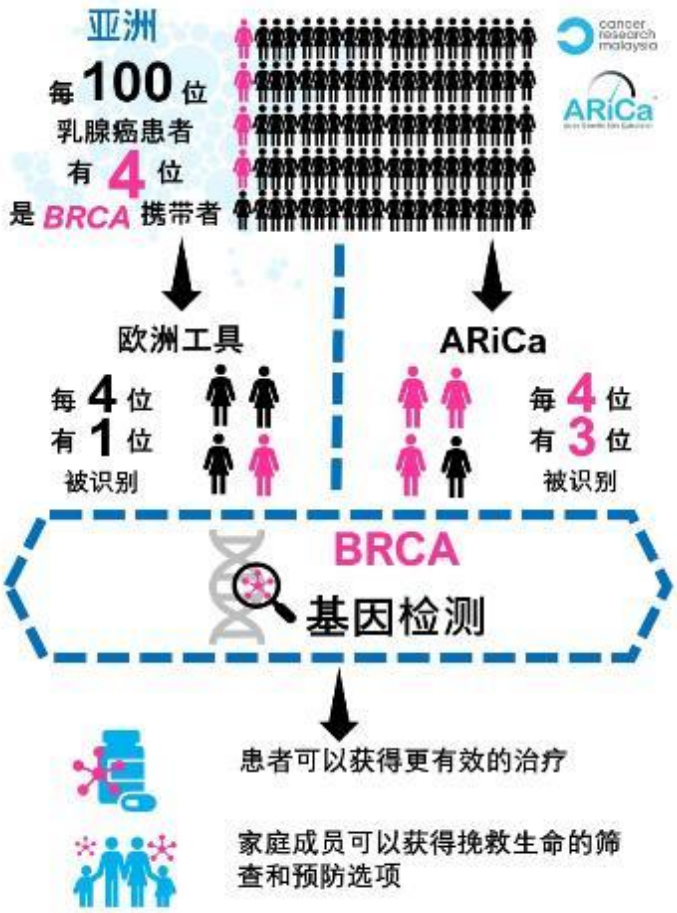
该项研究的完成有赖于来自惠康基金会, 森那美基金会, 国油基金会, 雅诗兰黛旗下公司, 机兴海星基金会, Malaysian Ministry of Higher Education High Impact Research Grant, 新加坡国立研究基金会, 新加坡国立大学, 国家医学研究理事会, 以及剑桥大学英国癌症研究协会的研究资助和慈善基金的支持。

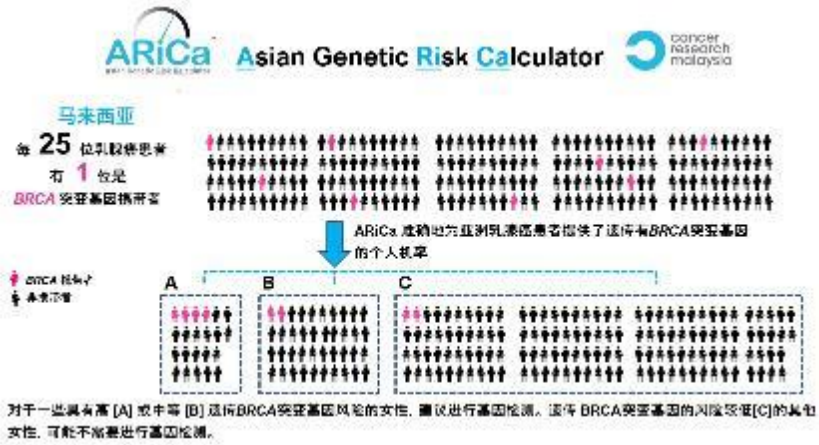
马来西亚癌症研究基金会博后科学家洪文凤博士表示："作为本研究的第一作者，我十分荣幸能够将该研究发表在一个具有高影响力的期刊。这成就有赖于马来西亚乳癌基因研究(MyBrCa)、新加坡乳癌队列研究(SGBCC)、以及许多在本地和国际层面上的长期合作。"

阅读该项研究: https://ascopubs.org/doi/abs/10.1200/JCO.21.01647

了解更**关于** BRCA 检测：www.embracingbrca.cancerresearch.my

获取基因检测：http://www.embracingbrca.cancerresearch.my/ 或联络 012 374 7426 or 016 363 4742





对于一些具有高 [A] 或中等 [B] 遗传BRCA突变基因风险的女性，建议进行基因检测。遗传 BRCA突变基因的风险较低[C]的其他女性，可能不需要进行基因检测。

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About Cancer Research Malaysia

Cancer Research Malaysia is the only non-profit organisation in Malaysia dedicated to saving lives through impactful research focusing on the Malaysian population and communities across Asia. Our research has already led to the discovery and implementation of new and effective breast cancer prevention strategies and our priority is ensuring that Asians are not left out in the fight against cancer. Together with our partners and supporters, Cancer Research Malaysia's vision is a future free of the fear of cancer. Funding for our lifesaving research depends on donations and sponsorship from the public and corporations. For more information, please visit cancerresearch.my or follow us on Facebook, LinkedIn, Instagram or Twitter.

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About the University of Cambridge

The mission of the University of Cambridge is to contribute to society through the pursuit of education, learning and research at the highest international levels of excellence. To date, 109 affiliates of the University have won the Nobel Prize.

Founded in 1209, the University comprises 31 autonomous Colleges, which admit undergraduates and provide small-group tuition, and 150 departments, faculties and institutions. Cambridge is a global university. Its 19, 000 student body includes 3,700 international students from 120 countries. Cambridge researchers collaborate with colleagues worldwide, and the University has established larger-scale partnerships in Asia, Africa and America.

The University sits at the heart of the 'Cambridge cluster', which employs 60,000 people and has in excess of £12 billion in turnover generated annually by the 4,700 knowledge-intensive firms in and around the city. The city publishes 341 patents per 100,000 residents.
www.cam.ac.uk

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About Universiti Malaya

Situated in the southwest of Kuala Lumpur, Universiti Malaya (UM) is the first university in Malaysia. UM is being supported by two academies, thirteen faculties, three institutes and three academic centres that comprehensively encompass medicine, science, technology, social sciences and humanities. UM also has the first and biggest teaching hospital in Malaysia, which is the University of Malaya Medical Centre (UMMC). The core of UM's contributions to the academia and society is through teaching, research, publication, innovation, and commercialisation.

UM has emerged among the world's top 60 universities and is ranked 59th on the Quacquarelli Symonds (QS) World University Rankings 2021. Since its establishment, UM has successfully produced approximately 200,000 graduates. For more information, please visit www.um.edu.my.

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About Subang Jaya Medical Centre

Subang Jaya Medical Centre is the flagship of Ramsay Sime Darby Health Care, a joint venture between Ramsay Health Care, Australia and Sime Darby. It is a licensed 444-bed multi-disciplinary and tertiary care private hospital nestled in the busting municipality of Subang Jaya, about 30 minutes' drive to Kuala Lumpur city centre and the Kuala Lumpur International Airport via major highways. The Hospital was established in 1985 and for decades, has provided comprehensive and complex care in all specialties. SJMC is also a tertiary referral hospital, receiving local patient referrals from within Malaysia as well as international patients from the Asia-Pacific region, in addition to serving as a major health care provider to a population catchment of an estimated 6.47 million.

About University of Nottingham

The University of Nottingham is a research-intensive university with a proud heritage, consistently ranked among the world's top 100. Studying at the University of Nottingham is a life-changing experience and we pride ourselves on unlocking the potential of our students. We have a pioneering spirit, expressed in the vision of our founder Sir Jesse Boot, which has seen us lead the way in establishing campuses in China and Malaysia - part of a globally connected network of education, research and industrial engagement. The University's state-of-the-art facilities and inclusive and disability sport provision is reflected in its status as The Times and Sunday Times Good University Guide 2021 Sports University of the Year. We are ranked eighth for research power in the UK according to REF 2014. We have six beacons of research excellence helping to transform lives and change the world; we are also a major employer and industry partner - locally and globally. Alongside Nottingham Trent University, we lead the Universities for Nottingham initiative, a pioneering collaboration which brings together the combined strength and civic missions of Nottingham's two world-class universities and is working with local communities and partners to aid recovery and renewal following the COVID-19 pandemic.

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About the National University Health System (NUHS)

The National University Health System (NUHS) aims to transform how illness is prevented and managed by discovering causes of disease, development of more effective treatments through collaborative multidisciplinary research and clinical trials, and creation of better technologies and care delivery systems in partnership with others who share the same values and vision.

Institutions in the NUHS Group include the National University Hospital, Ng Teng Fong General Hospital, Jurong Community Hospital and Alexandra Hospital; three National Specialty Centres - National University Cancer Institute, Singapore (NCIS), National University Heart Centre, Singapore (NUHCS) and National University Centre for Oral Health, Singapore (NUCOHS); the National University Polyclinics (NUP); Jurong Medical Centre; and three NUS health sciences schools – NUS Yong Loo Lin School of Medicine (including the Alice Lee Centre for Nursing Studies), NUS Faculty of Dentistry and NUS Saw Swee Hock School of Public Health.

With member institutions under a common governance structure, NUHS creates synergies for the advancement of health by integrating patient care, health science education and biomedical research.

As a Regional Health System, NUHS works closely with health and social care partners across Singapore to develop and implement programmes that contribute to a healthy and engaged population in the Western part of Singapore.

For more information, please visit http://www.nuhs.edu.sg.

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About A*STAR's Genome Institute of Singapore (GIS)

The Genome Institute of Singapore (GIS) is an institute of the Agency for Science, Technology and Research (A*STAR). It has a global vision that seeks to use genomic sciences to achieve extraordinary improvements in human health and public prosperity. Established in 2000 as a centre for genomic discovery, the GIS will pursue the integration of technology, genetics and biology towards academic, economic and societal impact.

The key research areas at the GIS include Human Genetics, Infectious Diseases, Cancer Therapeutics and Stratified Oncology, Stem Cell and Regenerative Biology, Cancer Stem Cell Biology, Computational and Systems Biology, and Translational Research.

The genomics infrastructure at the GIS is utilised to train new scientific talent, to function as a bridge for academic and industrial research, and to explore scientific questions of high impact.

For more information about GIS, please visit www.a-star.edu.sg/gis.

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