完善罕见病医疗保障体系，助力实现全民健康战略目标

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摘要

党的二十届四中全会通过的《中共中央关于制定国民经济和社会发展第十五个五年规划的建议》作出了“加快建设健康中国”的重大战略部署。习近平总书记强调：“人民健康是社会主义现代化的重要标志”[2]。世界卫生大会已将罕见病确立为全球健康优先事项，罕见病问题因此日益受到国际社会的广泛关注[3]。中国现有罕见病患者总数估计超2000万人[4]，且由于疾病复杂、诊断困难、治疗匮乏，部分患者仍承受着沉重的健康与经济负担。在“十五五”时期推进健康中国建设，需要为罕见病患者提供更为公平可及、优质高效、可负担的医疗卫生服务。同时，提升罕见病高值药物的支付能力，更能够破解阻碍创新药发展的关键瓶颈，促进生物医药战略性新兴产业发展，从而在“十五五”时期构筑产业发展新优势。

中国近年来通过国家多部委综合施策，在罕见病防治与保障方面已经取得了一定成果，已初步建成以基本医保为主体，医疗救助为托底，补充医疗保险、商业健康保险、慈善捐赠、医疗互助等共同发展的多层次医疗保障体

[1] 本报告仅代表企业相关研究观点，不代表论坛主办单位和承办单位立场和观点。

[2] 新华网. “习近平在福建考察时强调 在服务和融入新发展格局上展现更大作为 奋力谱写全面建设社会主义现代化国家福建篇章”. 2021年3月25日 https://www.xinhuanet.com/politics/leaders/2021-03/25/c_1127254519.htm

[3] 世界卫生组织. “罕见病: 全球卫生公平和包容的优先事项.” 2025年5月27日. https://apps.who.int/gb/ebwha/pdf_files/WHA78/A78_R11-ch.pdf.

[4] 病痛挑战基金会, 沙利文. “2023中国罕见病行业趋势观察报告.” 2023. https://www.frostchina.com/content/insight/detail/63f8f07f9c446c956d9754e3.

系。然而，现阶段多层次医疗保障体系在罕见病领域仍存在明显缺口，亟需在国家层面统一构建系统性制度安排，以进一步提升罕见病患者的整体保障能力。具体而言，基本医保由于其“保基本”的定位，对高值罕见病药物保障能力有限，且由于各地基本医保待遇差异，部分罕见病患者仍然面临用药“最后一公里”障碍。商业健康险由于其风险控制的商业逻辑，无法成为罕见病保障的有效补充手段，而慈善捐赠受自身筹资来源和运行模式限制，难以提供稳定可持续的医疗保障。

纵观全球各国罕见病保障体系，建立罕见病专项保障基金以统筹高值罕见病药品的支付，是中国在完善罕见病多层次医疗保障体系、提升高值药物支付能力方面可重点借鉴的重要模式。随着社会经济发展水平不断提高，中国已基本具备建立罕见病专项基金的经济基础。近年来，国内部分经济发达地区已率先开展罕见病专项基金的实践探索，充分验证了该模式在中国的可行性。

结合各国罕见病保障实践、国内地方试点成果以及中国罕见病多层次保障体系的现实基础，建议采取“国家统筹设计、地方试点探索”相结合的模式，逐步推进建立国家罕见病专项基金。在国家层面完成专项基金的顶层设计与筹备工作的同时，可在罕见病防治体系较为完善的地区率先开展试点。在国家统一指导下，充分发挥地方先行先试的优势，为专项基金的全面推广和全国覆盖奠定坚实基础。

（一）结合国际主流做法、国内地方试点成果以及中国多层次医疗保障体系的现实基础，国家罕见病专项基金的设计与实施可重点从以下方面展开：

- 专项基金需明确负责全国基金监督、管理、执行的部门职责，并对筹资、准入、支付、结算、评估等关键环节做出系统性制度安排，构建“可监管、可持续、可追溯”的基金管理模式。

- 专项基金需建立可持续的筹资来源，可综合考虑中央财政拨款、彩票公益金收入、慈善组织捐助等多元渠道。所需资金总量则可基于罕见病目录所涵盖疾病种类和国家罕见病直报系统中所登记患者基数开展测算。

- 国家可组织专家委员会，对拟纳入专项基金保障范围的药品进行系统评估和遴选，并明确优先保障的药品品种和罕见病类型。在药品价值评估方面，可构建更为灵活的价值评估体系，以有效应对罕见病由于患者规模较小所带来的价值评估挑战。

- 专项基金可同企业签订创新支付协议，采用基于财务结果的支付模式，构建高值药物风险共担机制，并设定个人自付上限。在将患者负担控制在可承受范围内的同时，这一机制还能提升专项基金支出的可控性与可持续性。

- 专项基金需建立同基本医保相衔接的机制安排，实现多层次医疗保障制度之间的协同互补，为罕见病患者提供覆盖全生命周期的连续保障。专项基金可在创新支付协议期间收集的真实世界数据，并基于相关数据对药品临床价值和基金支出影响开展评估，为相关药品参与医保谈判提供证据支持。

（二）建议在国家统一指导下，优先选择罕见病诊疗防治体系较为完善的地区开展专项基金试点，为全国推广积累可复制、可推广的实践经验

根据以往医保改革的经验，在国家方案制定的同时开展地方探索，有助于提前发现问题并积累实践经验。因此，专项基金在国家层面设计过程中，可在诊疗能力较强的地区率先开展试点，如开展基金规模测算、探索真实世界数据收集、试点签订创新支付协议等。通过地方探索验证提升国家方案设

计的科学性与可行性，确保专项基金制度既契合中国国情，也能够真实反映中国罕见病诊疗的客观实践。

一、引言

1.“十五五”时期建设健康中国需要提升罕见病患者医疗卫生服务水平

党的二十届四中全会通过的《中共中央关于制定国民经济和社会发展第十五个五年规划的建议》(以下简称《建议》) 做出了“加快建设健康中国”的重大战略部署。习近平总书记强调：“人民健康是社会主义现代化的重要标志”[5]。罕见病是威胁人类健康的重大公共卫生挑战。全球已知罕见病超过 7000 种，其中约 80%具有遗传性[6]。中国已确认的罕见病超过 1400 种，虽然单病种患病率极低，但由于中国人口基数大，现有罕见病患者总数估计超 2000 万人[7]。且由于疾病复杂、诊断困难、治疗匮乏，罕见病患者普遍承受着沉重的健康与经济负担。罕见病对儿童健康影响尤为突出，全球范围内 70%的罕见病在儿童期发病[8]，中国罕见病患者中 36.07%的确诊病例患者年龄＜18 岁[9]。改善罕见病患者的健康水平有助于改善儿童整体健康水平、提升为全人群提供公平可及的健康服务的能力，从而推动“健康中国”目标的实现并促进人口高质量发展。

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[5] 新华网. “习近平在福建考察时强调 在服务和融入新发展格局上展现更大作为 奋力谱写全面建设社会主义现代化国家福建篇章”. 2021 年 3 月 25 日 https://www.xinhuanet.com/politics/leaders/2021-03/25/c_1127254519.htm

[6] The Lancet Global Health. “The Landscape for Rare Diseases in 2024.” *The Lancet Global Health,* vol. 12, no. 3, 2024, e341. https://doi.org/10.1016/S2214-109X(24)00056-1.

[7] 病痛挑战基金会, 沙利文. “2023 中国罕见病行业趋势观察报告.” 2023. https://www.frostchina.com/content/insight/detail/63f8f07f9c446c956d9754e3.

[8] The Lancet Global Health. “The Landscape for Rare Diseases in 2024.”

[9] Guo, Jian, Peng Liu, Limeng Chen, et al. “National Rare Diseases Registry System (NRDRS): China’s First Nation-Wide Rare Diseases Demographic Analyses.” Orphanet Journal of Rare Diseases vol. 16, no. 1, 2021, p. 515. https://doi.org/10.1186/s13023-021-02130-7.

2.“十五五”时期完善罕见病保障体系能够在卫生领域展现中国的制度优势

近年来，国际社会日益重视罕见病对于全球健康所造成的挑战，将罕见病纳入全球卫生议程。2025 年召开的第 78 届世界卫生大会（WHA）通过了《罕见病：全球卫生公平和包容的优先事项》决议（以下简称《WHA 决议》），首次将罕见病确立为全球健康优先事项，并提出“实现包括罕见病患者在内的全民健康覆盖”的目标[10]。中国作为该决议的发起方之一，近年来通过国家多部委综合施策，在罕见病防治与保障方面已经取得了一定成果。随着《建议》明确提出“扩大高水平对外开放”“提供更多国际公共产品”的目标，中国将进一步深入参与引领全球卫生治理。完善罕见病医疗保障体系的中国方案，不仅是中国积极响应《WHA 决议》的重要举措，更可为全球罕见病事业贡献“中国智慧”。

3. 中国罕见病防治与保障事业已经取得显著成就

目前中国的罕见病防治与保障事业已经取得显著成就，国家在医疗、医保、医药领域推出了一系列政策措施，全面提升罕见病诊疗与保障能力。在罕见病药品领域，国家药品监督管理局通过优化审评审批激励罕见病新药研发，并且通过建立临床急需进口通道，加快引进国际上在研或在产的罕见病药物。在罕见病诊疗管理领域，国家卫生健康委开展系统行动，从出台罕见病目录、明确诊疗技术规范、到组织诊疗协作网、构建数据平台，形成了罕见病诊疗的“诊断、质量、保障、管理和研究”全链条方案。国家医疗保障局则初步建立了以基本医保、大病保险、医疗救助为主体的三重保障体系，

[10] 世界卫生组织. “罕见病: 全球卫生公平和包容的优先事项.” 2025 年 5 月 27 日. https://apps.who.int/gb/ebwha/pdf_files/WHA78/A78_R11-ch.pdf.

通过将罕见病药物纳入国家医保目录，并探索建立商业健康保险创新药目录，综合提升罕见病患者的保障水平。

二、完善罕见病药品保障体系对于“十五五”时期中国生物医药产业创新发展具有重要意义

罕见病诊疗能力的提升离不开创新药的发展。由于约有 95%的罕见病仍缺少有效治疗方案，近年来罕见病已成为全球新药研发的热点领域之一[11]。据贝哲斯咨询统计，2025 年全球孤儿药市场规模达 15455.89 亿元人民币[12]。对罕见病发病机制和创新疗法的研究，更可推动常见病治疗技术创新，具有巨大的科技和经济价值。例如，降血脂新药 PCSK9 抑制剂的发现即来源于针对罕见基因突变的研究[13]。

罕见病领域也是中国生物医药产业创新发展的高潜力领域之一。中国人口基数大、罕见病患者招募快、临床研究成本低、人工智能发展迅速，在罕见病药物研发领域具有独特优势，有望在罕见病领域实现创新药发展“弯道超车”。在罕见病药物研发和全链条支持创新药发展等政策的支持下，中国罕见病药物创新发展迅速，研发能力和临床价值得到国际认可。2024 年，FDA 共向 481 种药物授予孤儿药认证，其中 68 种由中国药企所研发[14]。从创新药物发源国角度来看，中国位列第二。中国已成为全球生物医药创新的重要力量之一。

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[11] 张抒扬, 张学. “近年中国罕见病相关政策和实践探索.” 罕见病研究, vol. 1, no. 1, 2022, pp. 1–6. https://doi.org/10.12376/j.issn.2097-0501.2022.01.001.

[12] 贝哲斯咨询. “2025 年孤儿药市场发展趋势报告：容量、价格走势及竞争调研.” 2025 年 8 月 4 日. https://www.sohu.com/a/929318840_122448164.

[13] 封思琴, 王一斐, 陈沛沛, 张志宇 , 张抒扬. “PCSK9 抑制剂多效性的研究进展.” 中华心血管病杂志, vol. 50, no. 3, 2022. https://csc.cma.org.cn/art/2022/7/6/art_620_45976.html.

[14] 药时代. “481 款! 2024 年 FDA 孤儿药认证大爆发! 中国排名第二.” 2025 年 3 月 25 日. https://pharm.jgvogel.cn/c1498564.shtml.

目前，中国创新药发展仍然面临创新药企价格期待与医保支付能力尚有落差、多元化支付能力薄弱等问题[15]。定价与支付政策已经成为本土罕见病药物从创新研发走向产业转化并拓展国际市场所面临的主要制约因素。《建议》确立了“全面增强自主创新能力，不断催生新质生产力”的发展战略。《建议》同时明确指出，“十五五”时期将“加快建设健康中国”，并将“支持创新药和医疗器械发展”作为推进健康中国建设的举措之一。因此，完善罕见病医疗保障体系，对于完善创新药生态体系建设，促进生物医药战略性新兴产业发展，在“十五五”时期构筑产业发展新优势具有重要意义。

本报告基于中国罕见病保障体系的发展现状与挑战，系统梳理国际经验与国内实践，并在此基础上提出设立国家罕见病专项保障基金的政策建议，旨在为中国在“十五五”时期构建更加公平、高效、可持续的罕见病保障体系，推动医药创新发展与全民健康目标的实现提供决策参考。

三、中国罕见病医疗保障体系建设进展及挑战

（一）罕见病医疗保障体系建设进展：初步建立多层次保障体系

目前中国的罕见病防治与保障事业已取得显著成就，初步建成以基本医保为主体，医疗救助为托底，补充医疗保险、商业健康保险、慈善捐赠、医疗互助等共同发展的多层次医疗保障体系。

1. 国家医保药品目录通过谈判纳入罕见病药品

自 2017 年起，国家医保局等相关部门开始通过医保谈判的方式将罕见病药物纳入国家医保药品目录，有效提升了罕见病药物的保障水平。截至 2024 年底，已有 126 种罕见病药品被纳入国家医保药品目录，覆盖 68 个罕

[15] 国家医疗保障局. “国家医保局“支持创新药高质量发展的若干措施”新闻发布会实录.” 2025 年 7 月 1 日. https://www.nhsa.gov.cn/art/2025/7/1/art_14_17065.html.

见病种类，约占已上市罕见病药品的三分之二[16]。2025 年基本医保目录更新持续扩展罕见病药物覆盖，将 10 种罕见病用药新增纳入基本医保目录，进一步提升罕见病保障水平[17]。

2. 地方通过大病医保等方式覆盖部分不在目录内的药品

针对未纳入国家医保药品目录的罕见病，地方政府积极探索通过设立专项基金、扩展大病保险覆盖范围等模式，为罕见病患者提供补充保障，一定程度上缓解了部分罕见病患者的疾病负担。浙江省通过设立罕见病专项基金，将戈谢病、庞贝病、法布雷病和苯丙酮尿症患者纳入保障范围，为患者使用未纳入国家医保药品目录的高值罕见病创新药提供保障。湖南省则将未能进入国家医保药品目录、用于戈谢病和庞贝病治疗的两种药品，纳入了当地大病医保报销范围。通过谈判和企业分担药品成本，由大病医保承担 50%的药品费用，企业承担约 40%，将个人自付费用控制在 3 万元以内。

3. 惠民保为罕见病患者提供衔接基本医保的补充保障

由地方政府主导，保险企业承保的普惠补充商业健康保险也为罕见病患者提供了一定补充保障。截至 2025 年 7 月 31 日，全国累计推出 313 款地方性惠民保产品，其中正常运营产品数量为 202 款[18]。近 3 年超 50%的惠民保产品已纳入罕见病用药[19]。大部分惠民保产品会对国家医保药品目录内罕见

[16] 病痛挑战基金会, 沙利文. “2025 中国罕见病行业趋势观察报告.” 2025 年 2 月 28 日. https://www.frostchina.com/content/insight/detail/67bed1ec7ed30cc08c184b97.

[17] 经济日报. “医保商保‘双目录’大力支持创新——好药新药加速惠及百姓.” 2026 年 1 月 3 日. https://www.news.cn/fortune/20260103/a458cb7c27b3499c823fc43b29777a3e/c.html.

[18] 每日经济新闻. “价格涨了, 产品少了, ‘基本盘’稳了 惠民保进入‘成熟期’.”. 2025 年 10 月 22 日. https://www.nbd.com.cn/articles/2025-10-22/4101569.html.

[19] 健康报，“罕见病用药保障如何再进一步——对话中国药科大学国际医药商学院副院长茅宁莹”. 2025 年 11 月 14 日，https://www.jkb.com.cn/horizon/2025/1114/507068.html

病药品的患者自付费用进行再次报销，或将国家医保药品目录外的高值罕见病创新药纳入报销范畴[20]。

2025 年 12 月，国家医保局公布首版商业健康保险创新药品目录，共纳入 19 种药品，其中 6 种为治疗神经母细胞瘤、戈谢病、苯丙酮尿症的罕见病药品，为推动商业健康保险与基本医保错位发展，进一步完善多层次医疗保障体系奠定了基础[21]。

4. 医疗救助与慈善力量为罕见病患者提供托底保障

各地通过医疗救助为罕见病患者设立专项补助，为患者和其家庭可能面临的灾难性医疗支出提供托底保障，降低因病致贫或返贫的风险。以佛山市为例，佛山将戈谢病、法布雷病、糖原累积病（II 型）等 50 余种罕见病药品和治疗性食品费用纳入了医疗救助范围，对符合条件的救助对象，经基本医保、大病保险和各类补充医疗保险核报后，个人先行支付剩余自付费用，再由户籍所在区医保部门按 80%的救助比例予以救助，年度救助限额为 30 万元。

地方慈善公益力量则发挥其个性化、多元化服务能力，为罕见病患者提供援助支持。佛山市医疗保障协会、佛山市妇幼保健院与病痛挑战基金会三方在佛山市医保局的指导下，于 2023 年启动了佛山市罕见病慈善医疗救助和综合服务试点项目，打造了全国第一个市域罕见病医疗保障、慈善医疗救助、综合服务一体化样板。由病痛挑战基金会在佛山妇幼保健院派驻罕见病医务社工，提供患者筛查、救助申请、用药支持和心理辅导服务，并将佛山市罕见病医疗救助流程嵌入医疗机构的诊疗与社会工作流程。病痛挑战基金

[20] 健康报. “罕见病用药保障如何再进一步.”. 2025 年 11 月 14 日. https://www.jkb.com.cn/horizon/2025/1114/507068.html.

[21] 第一财经. “6 款罕见病药物纳入首版商保创新药目录, 如何吸引更多商保承接.” 2025 年 12 月 10 日. https://news.qq.com/rain/a/20251210A05VUO00.

会的项目执行团队还定期协助佛山市医疗保障协会核对所有患者的医保支付数据，避免重复资助。

（二）现有体系的主要挑战和局限性

尽管中国已初步构建多层次的罕见病医疗保障体系，但整体保障能力仍有进一步提升的空间，尤其是在高值罕见病药物的支付保障方面，仍需不断完善。研究显示[22]，中国约有 1.1%的罕见病患者面临极高的疾病负担[23]，这类患者自付的直接医疗费用占其家庭年收入的平均比例高达 2460.81%。而对于约占 70.2%的高负担罕见病患者[24]，其自付直接医疗费用占家庭年收入的比例也已达到 95.88%[25]。考虑到约 60%的罕见病患者家庭年收入低于 3 万元[26]，罕见病的“家庭灾难性支出”极易造成患者家庭因病致贫或因病返贫。因此，探索建立中国的高值罕见病药物支付保障机制，是目前中国罕见病患者群体最迫切的需求。

1.基本医保的“保基本”定位限制了对高值药物的支付能力

罕见病药品研发成本高、周期长，但由于罕见病患者人数稀少，使得罕见病药品定价普遍较高。目前仍有 65 种药物未被纳入基本医保，涉及 58 种罕见病，其中全部治疗药物均未纳入基本医保且费用高昂的药物有 19 种，涉及 26 种罕见病[27]。对于基本医疗保险尚未覆盖的罕见病患者，绝大多数都

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[22] Yu J, Chen S, Zhang H, et al. “Patterns of the health and economic burden of 33 rare diseases in China: nationwide web-based study.” *JMIR public health and surveillance*, 2024, 10: e57353.

[23] 极高疾病负担罕见病患者主要特征为个体报告中度至重度身体和心理健康问题的可能性显著增加，同时伴随直接医疗、直接非医疗以及间接费用相对于家庭收入而言的平均自付医疗支出水平显著偏高。

[24] 高负担罕见病患者的主要特征为参与者报告中度至重度健康问题的概率较高，同时疾病相关的平均经济支出相对于家庭收入而言也处于较高水平。

[25] Yu J, Chen S, Zhang H, et al. “Patterns of the health and economic burden of 33 rare diseases in China: nationwide web-based study.”

[26] 央广网. “2025 听两会丨全国人大代表吴焕淦: 建立罕见病多层次保障体系” 2025 年 3 月 10 日. https://roll.sohu.com/a/869005026_362042.

[27] 病痛挑战基金会, 沙利文. “2025 中国罕见病行业趋势观察报告.”

因为高昂的费用选择了放弃治疗，或选择非常有限的“不足量不足疗程”的用药方式，这些都将严重影响患者的生命健康[28]。

2.用药“最后一公里”困境：基本医保待遇差异导致部分患者仍面临沉重的自付负担

尽管部分罕见病药品已经纳入国家医保目录，由于各地基本医保待遇差异，部分罕见病患者仍然面临用药“最后一公里”的障碍。很多罕见病需要长期在门诊进行治疗和随访，一项关于中国 300 个地级市门诊慢性病和特殊病种纳入情况的研究显示，只有 24 种罕见病被地方医保覆盖，且大多数城市覆盖的罕见病少于 4 种，因此很多罕见病患者仍无法在门诊获得治疗并享受报销待遇[29]。此外，各地对国家医保药品目录内罕见病药品的门诊报销比例也存在显著差异，且城乡居民基本医保的门诊报销比例相对较低，部分省市的门诊医保无法衔接大病医保，导致罕见病患者自费负担仍然比较沉重。

3.商业化运作的健康保险难以作为罕见病保障的有效补充手段

目前中国的商业健康险覆盖率处于较低水平，健康险深度仅为 0.7%，远低于发达国家水平[30]。作为市场化的医疗风险分担机制，商业健康保险运用大数法则，将小概率的个体健康风险通过扩大参保人群实现风险分散[31]。由于约 80%的罕见病具有遗传性，罕见病群体中高风险个体比例明显偏高。为了避免健康人群参保意愿不强、高风险带病人群积极参保，导致赔付率超

[28] *Ibid*.
[29] Xu, Juan, Mingren Yu, Zhiguo Zhang, Shiwei Gong, Bingqin Li. “Is Sub-National Healthcare Social Protection Sufficient for Protecting Rare Disease Patients? The Case of China.” Front. Public Health, 2023;11.
[30] 财中社. “国联证券：我国商业健康险实际覆盖率较低.” 2025 年 1 月 17 日. https://news.qq.com/rain/a/20250117A029QM00.
[31] “商业健康保险在中国罕见病医疗保障中的应用现状、问题及对策.” 罕见病学杂志. 2022 年 1 月 14 日. https://jrd.chard.org.cn/cn/article/doi/10.12376/j.issn.2097-0501.2022.01.014.

出预期的“逆向选择”风险，商业健康保险往往需要设置既往症限制等方式控制风险，因此无法为罕见病患者提供有效的补充保障。

而由地方政府主导的普惠补充商业健康保险目前仍处于市场调整阶段，产品设计处于不断优化与迭代的过程中。目前，不同地区的“惠民保”产品设计存在较大差异，各产品在保障范围和待遇水平上并不一致，罕见病患者能够获得的保障水平亦因此不同。截至 2025 年 7 月，47 个“惠民保”产品收录有罕见病，占所有惠民保产品的 25%，收录有罕见病的惠民保产品分布在 42 个城市，占所有销售惠民保产品城市的 30%[32]。对于罕见病患者而言，这意味着“惠民保”目前尚不能作为一项稳定可靠的补充保障手段，部分罕见病患者仍可能因保障不足而面临较为沉重的疾病负担。

4. 慈善捐赠无法作为罕见病患者的主要保障来源

慈善力量是罕见病用药保障“最后一公里”的重要补充，但受其自身属性限制，难以成为稳定可持续的核心保障来源。一是资金来源不稳定。据统计，超 60%以上慈善资金依赖企业捐赠，受社会关注度与经济环境影响大；且仅有约 18%的捐赠意向为医疗健康领域[33]。有调查结果显示，公众对罕见病认知很有限，约有 75%的公众知道 3 种及以下的罕见病，极大的限制了慈善捐赠对于罕见病的支持能力[34]。二是保障覆盖碎片化。慈善项目多针对特定病种，覆盖范围、申请标准不统一。参与重特大疾病的公益慈善项目主要关注恶性肿瘤等重大疾病，投入罕见病领域仅占捐赠额 5%[35]。三是罕见病多需长期甚至终身治疗，但多数慈善项目资助期仅 1 年且设有资金限额，短

[32] 和观医疗. “商保创新药申报之: 罕见病治疗药的候选名单及现有惠民保准入分布.” 2025 年 7 月 2 日. https://mp.weixin.qq.com/s/L8T1K5ILOUbDRA4NrQ9ODg.

[33] 宋宗合. “2019-2020 年度中国慈善捐赠报告载杨团.” 载*《慈善蓝皮书：中国慈善发展报告 2021》*杨团, 朱健刚主编. *社会科学文献出版社*. 2021 年 12 月.

[34] 深圳国际公益学院。《罕见病公众认知调查报告》发布，联动多方助力解决用药困境 2023 年 2 月 28 日.https://www.cgpi.org.cn/index/hotnewstext/id/1335.html

[35] Deloitte, 患者援助联合行动同盟. “2021 年中国患者援助年鉴.” 2021.

期化、定额化的资助模式，无法匹配患者长期用药的刚性需求。可见，慈善力量在罕见病保障体系中更多扮演填补空白、缓解急难问题的角色，而非承担系统性支付责任。

综上，目前中国多层次医疗保障体系在罕见病保障方面仍存在一定缺口，且罕见病患者的保障水平在不同地区之间存在明显差异。因此，亟需在国家层面统一构建具有可持续支付能力的制度安排，以系统性提升中国罕见病领域的保障水平、为创新药发展提供持续动力。

四、国际和国内政策经验

纵观全球各国的罕见病保障体系，主要模式包括以专项基金为主的多元共付模式、以全民健康保险为主的保障模式、和以商业保险为主的保障模式。

- 法国采取了由法定医保基金主导的保障模式。法国建立了长期性疾病（Affections de Longue Duree，ALD）清单制度，ALD30 清单中包括部分罕见病，对于 ALD 30 所包含的疾病，其医疗费用可得到 100%报销[36]。同时，对于 ALD30 未包含，但治疗需超过 6 个月且费用昂贵的疾病，患者可个案申请医保 100%报销。
- 在美国，绝大多数人群的医疗保障由商业保险提供，而低收入群体和老年人群体则分别由医疗补助计划（Medicaid）和联邦医疗保险计划（Medicare）提供保障。《平价医疗法案》（Affordable Care Act）规定绝大多数商业保险计划不得因既往病史（包括罕见病）拒绝承保[37]，但该法案并未要求明确哪些罕见病疗法必须

[36] Directorate of Legal and Administrative Information (Prime Minister). “Management of a long-term illness (ALD) by the Health Insurance.” 获取日期 2026 年 3 月 10 日 https://www.service-public.gouv.fr/particuliers/vosdroits/F34068?lang=en.

[37] U.S. Department of Health and Human Services. “Pre-Existing Conditions.” March 17, 2022. https://www.hhs.gov/healthcare/about-the-aca/pre-existing-conditions/index.html.

由商业健康保险报销，也未规定具体报销标准。对于联邦医疗保险计划（Medicare）而言，老年患者在达到“灾难性支出门槛”（2026 年为 2,100 美元）后，可获得高价处方药 95%的报销额度[38]。对于医疗补助计划（Medicaid），各州独立管理其可报销药品清单，因此对低收入患者孤儿药的覆盖范围存在州与州之间的显著差异[39]。

- 意大利则选择在全民健康保险之外建立罕见病专项基金，从而提高罕见病医疗费用保障能力[40]。意大利的罕见病专项基金利用了部分所向制药企业征收的药品推广税款和国家卫生基金拨款，为罕见病药物提供高达 100%的报销。

综合各国罕见病保障典型模式，并结合中国多层次医疗保障体系的现实基础，罕见病专项基金模式对于进一步优化中国罕见病多层次医疗保障体系具有重要的参考价值。

（一）各国罕见病专项基金的运营模式和管理经验主要呈现以下特点：

1. 基金设立有法律依据和稳定筹资来源

由于罕见病专项基金需要专门且可持续的筹资来源，通常来自国家税收收入和/或全民健康保险基金的拨付，因此很多国家都颁布了罕见病相关的法律法规，为罕见病专项基金的设立和运营提供了法律依据和制度保障。例如，2024 年，埃及修订了医疗应急基金相关法律，为罕见病患者的治疗提

[38] Wreschnig, Laura A. “Medicare Part D Prescription Drug Benefit.” *Congressional Research Service.* November 14, 2023. https://www.congress.gov/crs-product/R40611.
[39] KFF. “State Medicaid Drug Review Responsibilities.” July 1, 2019. https://www.kff.org/state-health-policy-data/state-indicator/medicaid-drug-review-responsibilities/.
[40] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. “The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy.” *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

供了制度化的财政保障[41]。法律规定，基金将负责支付特定罕见病药物和治疗的费用，使罕见病患者的治疗不再依赖零散项目、慈善组织或临时资金，从而形成长期且可持续的保障机制。

2. 以罕见病定义或罕见病目录作为专项基金保障病种的依据

虽然在全球层面并没有一个统一且被普遍接受的罕见病定义，各国普遍在罕见病相关立法或国家罕见病行动计划中都有对罕见病的明确定义，或者制定了国家罕见病目录，成为国家专项基金、全民健康保险或商业医疗保险确定罕见病保障病种的主要依据。以俄罗斯为例，对罕见病的定义是患病率不超过每 10 万人中 10 例（≤10/100,000）的疾病，同时俄罗斯联邦卫生部制定了罕见病目录，目前包括约 290 种疾病。在此基础上，负责管理 19 岁以下罕见病患者专项基金的俄罗斯“慈善圈”基金会，为该项目制定了专门的罕见病目录，至少每两年修订一次。自 2021 至 2025 年，该目录纳入的疾病数量已从 44 种增加至 101 种，共为超过 29000 名儿童提供了治疗药物。

3.覆盖国外已上市、国内未上市、国内已上市药物，鼓励创新药

为了保障罕见病患者能够及时获得创新疗法，各国在界定罕见病专项基金保障范围时会覆盖包括尚未在本国上市的药品和已经在本国上市的药品，从而最大程度提升创新疗法的可及性。以意大利为例[42]，其罕见病专项基金不仅保障欧洲药品管理局（EMA）已批准但尚未在意大利上市的孤儿药，也覆盖已在本国上市的药品。目前，EMA 批准的孤儿药中超过 80%都可以在意大利报销，而意大利的孤儿药支出与欧洲平均水平持平，且价格更低。

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[41] Tinnion, Violet. “Egypt Expands Rare Disease Funding through Emergency Medical Fund.” FrontierView, January 24, 2024. https://app.frontierview.com/insightBite/1881/egypt-expands-rare-disease-funding-through-emergency-medical-fund.

[42] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. “The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy.” *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

针对欧洲药品管理局已批准但尚未在意大利上市的孤儿药，意大利建立了“AIFA 5% 基金”为患者实际花费提供 100%报销。该基金来源为基于制药企业的药品推广支出所征收的 5%税款[43]，其中所获税款的 50%用于“AIFA 5%基金”。使用“AIFA 5% 基金”基金无需进行价格谈判，仅需医疗机构提交用药申请。

对于已经在意大利上市的药品，如被意大利药品监督管理局认定具备“完全创新性”（fully innovative），其上市后的 36 个月内的费用由“创新药物基金”100%报销[44]。意大利药品监督管理局每年认定的创新药物中约三分之一为孤儿药。“创新药物基金”的资金来源为国家卫生基金每年 13 亿欧元（约 107 亿元人民币）的拨款[45]。

4. 建立和企业风险共担创新支付协议，加速创新药可及并提升基金可持续性

各国的罕见病专项基金通过采取预算管理和风险共担协议等手段，帮助医保部门有效避免基金的透支风险，并为最需要治疗但又没有支付能力的患者提供保障，从而显著的减轻了罕见病患者家庭的医疗负担。风险共担模式是国际上已广泛应用于罕见病和抗肿瘤药物的一种创新支付机制，在促进新药快速进入临床使用的同时，提高医保基金使用效率、减少财务风险。以中国台湾地区为例，健保署于 2018 年 9 月将管理准入协议（Managed Entry Agreements，MEA，又译为“药品给付管理协议”）作为“其他协议方案”正式加入《全民健康保险药物给付项目及支付标准》，包含基于疗效结果的

[43] 自第 175/2021 号法令颁布起，税率已升至 7%，新增的 2%税款用于支持有关罕见病、易被忽视和先进疗法的公共研究。

[44] Vogler, Sabine. “Payer Policies to Support Innovation and Access to Medicines in the WHO European Region.” Copenhagen Ø, Denmark: World Health Organization. 2022. 4. Case studies. https://www.ncbi.nlm.nih.gov/books/NBK587872/.

[45] 2022 年之前为两个专项基金，分别用于创新肿瘤药物和创新非肿瘤药物的报销，2022 年合并为一个专项基金，当年的基金规模为 10 亿欧元。

协议方案和基于财务结果的协议方案两大协议方案类型[46]，协议期的上限为5 年，到期后可重新签约，且允许企业自行提出并和健保署商讨具体协议条件，按照协议应由企业返还的药费会并入患者的总额医疗费用进行结算。

5. 与国民健康保险实现有效衔接，为患者提供连续性保障，患者无需自付或自付部分在可负担范围内

在以专项基金或全民健康保险作为罕见病主要保障机制的发达国家和地区，政府通常对罕见病患者的药物费用实行全额（100%）报销。典型案例包括法国[47]、意大利[48]等国家。一些国家和地区的专项基金负责保障特定期限或阶段内的罕见病药物费用，同时通过构建与全民健康保险的衔接机制，为患者提供连续保障，有效降低其医疗费用的自付负担。例如，如前文所述，意大利药品监督管理局认定具备“完全创新性”的药品在上市后的 36 个月内，由“创新药物基金”100%报销，之后转由各大区在各自的医保预算范围内，按意大利药品监督管理局和企业谈判议定的价格进行报销。

在药品上市初期，意大利通过管理准入协议和与协议相对应的患者病例登记数据库，系统收集、评估药品使用数据，为未来的医保谈判价格和支付标准设定提供决策依据。意大利药品监督管理局批准报销的药品，大部分都签订了管理准入协议。作为和企业签订管理准入协议的配套措施，意大利自2005 年起建立了一个对应的全国性患者病例数据库，将协议药品的患者病例纳入数据库。截至 2019 年底，该数据库共录入了按适应症登记的 283 个

46 王美凤, 王海银, 丛鹂萱, 谢春艳, 刘昕, 金春林. 我国台湾地区创新药医保支付方式经验及启示研究. 世界临床药物, vol. 42, no. 3, 2024, pp. 223-228. https://doi.org/10.13683/j.wph.2021.03.014.

47 Service Public, Management of a long-term illness (ALD) by the Health Insurance, Accessed on March 5, 2026 https://www.service-public.gouv.fr/particuliers/vosdroits/F34068?lang=en

48 对于患有罕见或慢性疾病（包括艾滋病）的患者，以及孕妇，意大利免除其因病情相关治疗所需自付的费用。The Commonwealth Fund “International Health Care System Profiles: Italy”. Accessed on March 14, 2026. https://www.commonwealthfund.org/international-health-policy-center/countries/italy

病例，涉及 159 种药品，其中 88 个病例为罕见病，涉及 52 种孤儿药[49]。基于数据库所收集的信息，医保部门可以依据每个患者的疗效结果进行医保支付或要求企业返还药品费用，未来还可以进一步分析汇总后的数据，为选择最佳治疗方案提供数据支持。

（二）国内试点证明专项基金模式可行

近年来，国内一些经济发达地区也已率先开展探索，尝试建立面向罕见病的保障支付机制。其中，浙江省设立罕见病专项基金，试行“省级统筹、单独筹资”的模式。浙江的探索在筹资能力和保障水平方面均表现突出，专项基金运行稳定、支出均衡。浙江实践为国家层面建立罕见病专项基金提供了以下具有代表性的实践经验：

- **专款专用，确保稳定且可持续的资金来源：**每年每人 2 元一次性从大病保险基金中划转至省罕见病用药保障基金，并在省级医疗保险基金财政专户中下设子账户，进行分账管理、独立核算。
- **保障范围的病种包括戈谢病、庞贝病、法布雷病和苯丙酮尿症，通过专家论证确定保障病种：**专家委员会由罕见病临床医生、研究学者、医保政策专家等组成。
- **设定分段式报销比例和个人自付封顶线：**参保人员在一个结算年度内发生的药品费用，实行费用累加计算分段报销：0～30万元，报销比例为 80%；30～70 万元，报销比例为 90%；70 万元以上费用，全额予以报销，个人自付封顶线为 10 万元。

[49] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. “The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy.” *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

- **与医疗救助衔接：**经罕见病专项基金报销后的剩余费用，符合医疗救助条件的人员可向参保地政府申请获得医疗救助。
- **建立和企业的风险共担机制：**开展和企业的罕见病用药谈判，并设定高值罕见病药物的年度报销总费用上限。自设立以来，浙江省罕见病专项基金在 2020 年至 2024 年间连续保持年度结余，基金整体稳定运行。

五、政策建议

随着社会经济发展水平不断提高，中国已基本具备建立罕见病专项基金的经济能力。埃及 2024 年修订医疗应急基金相关法律、为罕见病患者的治疗提供了制度化的财政保障时，人均 GDP 为 3338 美元；俄罗斯 2021 年成立“慈善圈”基金会，用于支付 19 岁以下罕见病患者治疗费用时，人均 GDP 为 12425 美元[50]。“十四五”时期中国人均 GDP 从 1 万美元增至 1.3 万美元以上[51]，已位居中等偏上收入国家前列[52]。因此，中国应考虑从国家层面探索引入罕见病专项基金模式，进一步完善中国“1+3+N”多层次医疗保障体系，增强“N”其他保障力量对罕见病高值药物的支付能力，为罕见病患者提供可及可负担的医疗卫生服务。

建议采取“国家统筹设计、地方试点探索”相结合的模式，在国家层面完成国家罕见病专项基金的顶层设计与筹备工作。同时，在罕见病防治体系较为完善的地区开展专项基金试点，重点围绕筹资规模、创新支付机制、患者病例数据库等关键环节和关键流程进行探索，逐步积累可复制、可推广的

[50] World Bank Group. “World Bank Open Data: GDP per capita (current US$) - Russian Federation, Egypt, Arab Rep.” 2024. https://data.worldbank.org/indicator/NY.GDP.PCAP.CD?locations=RU-EG.
[51] 新华网. “十四届全国人大四次会议经济主题记者会.” 2026 年 3 月 6 日. https://www.news.cn/politics/20260306/81996586d4ac4e4faf95d0431089db68/c.html.
[52] 中国经济网.“国家统计局: 中国人均 GDP 已位居中等偏上收入国家前列.” 2025 年 10 月 1 日. http://www.ce.cn/xwzx/gnsz/gdxw/202510/t20251001_2501161.shtml.

实践经验和具体做法。在国家统一指导下，充分发挥地方先行先试的优势，为国家罕见病专项基金的全面推广和全国覆盖奠定坚实基础。

（一）结合国际主流做法、国内地方试点成果以及中国罕见病多层次保障体系的现实基础，国家罕见病专项基金的设计与实施可重点包括以下方面：

完善顶层设计，建立全流程管理机制：专项基金管理应采取多部门联合协作模式，设立全国基金的监督、管理、执行部门，并明确相关部门职责与部门分工。专项基金管理机制需对筹资机制、保障对象、准入标准、待遇水平、信息平台、管理办法、配套措施等做出系统性制度安排。遵循“可监管、可持续、可追溯”三项基本原则，建立覆盖筹资、准入、支付、结算、评估等关键环节的全流程管理机制，确保工作流程和人员可监管、筹资来源和规模可持续、资金使用和患者信息可追溯。

建立稳定可持续的筹资来源：由于中国基本医保基金收支平衡仍持续面临一定压力，而商业健康保险由于其商业逻辑需要通过扩大参保人群而分散风险，因此需要确定独立、稳定和可持续的筹资来源。建议综合考虑中央财政拨款、彩票公益金收入、慈善组织捐助等多元渠道，筹建国家罕见病专项基金池。

根据国家罕见病目录收录的疾病数量和国家罕见病直报系统登记的患者基数，以及尚未纳入国家基本医保药品目录和商业健康保险创新药品目录的药品数量和费用，同时设定固定的个人年度自付费用封顶线，可以初步测算出所需资金的总量。在此基础上，可以结合中央财政收支及预算分配情况，按照“以收定支、量力而行、循序渐进”的原则，决定国家罕见病专项基金的首期筹资规模和保障范围。

实行规范的申报评审机制，明确药品遴选标准：国家罕见病专项基金应主要将治疗国家罕见病目录内疾病、尚未纳入基本医保目录的罕见病药品纳入遴选范围。建议组建国家罕见病专项基金专家委员会，基于罕见病专用的价值评估体系，对企业申报的药物进行评估，并可考虑优先选择符合以下条件的药物：

- 治疗儿童罕见病的药物
- 治疗会危及生命或严重影响生活质量疾病的药物
- 能够解决未满足临床需求的创新药物
- 潜在长期获益和/或社会价值较高的药物

由于罕见病患病人数少，难以开展大规模的临床试验，也难以对药品上市前的临床试验数据进行准确的有效性评价，因此可以考虑采取更加灵活的价值评估体系，在准入评估时接受小样本研究或单臂研究的数据，并要求企业在准入后开展上市后真实世界研究，作为准入后价值评估和续约谈判的证据。

通过构建风险共担机制、引入创新支付协议管理，并设定个人自付上限：由国家罕见病专项基金提供保障的罕见病药物年治疗费用较高，因此需要参考国际、国内经验，结合临床治疗实际需求、专家委员会评估建议和国家罕见病专项基金的收支情况，组织和企业的创新支付协议谈判，构建风险共担机制。同时，可借鉴浙江经验，设定个人自付上限，不仅有效缓解罕见病药物高额治疗费用压力、将患者负担控制在可承受范围内，更可提升基金的可持续性。

对于患者基数明确、国内已上市、临床需求显著且疗效明确的罕见病特效药，建议价格谈判的基础上设定年度报销总费用上限，既便于专项基金相应预算和支出的管理，也能稳定企业的价格预期，确保药品的稳定供应。

对于患者基数尚不明确、临床疗效和对专项基金影响存在不确定性的罕见病新药，考虑到基于疗效结果的协议在研究设计、数据分析、患者返款等实际操作环节的难度较高，建议和企业签订基于财务结果的创新支付协议，明确意向支付价和风险分担方式，并对付款周期、比例和条件等进行详细约定。同时，建议参考意大利经验，和企业签订数据收集协议，明确协议执行各方的具体职责。企业可承担收集真实世界数据的相关费用。所获取的相关数据能够为药物在真实世界应用中的实际临床获益和风险提供有力证据[53]，从而支持该药品未来参与国家医保谈判。

建立与基本医疗保险的衔接机制，实现多层次医疗保障制度之间的协同互补：在现有的罕见病多层次保障体系中引入国家罕见病专项基金模式的同时，需要设计好其与基本医疗保险的衔接机制，实现对专项基金保障药品目录的动态管理和调出药品与国家基本医保药品目录的有效对接。通过实现专项基金保障与国家基本医保目录的有效对接，可为罕见病患者提供覆盖全生命周期的连续保障，从而持续降低整体疾病负担。

建议对首次纳入国家罕见病专项基金保障范围的药品，和企业签订为期不超过 5 年的创新支付协议，并同步启动真实世界数据收集工作，在协议期间对收集到的数据进行动态监管和期中分析。协议到期后，可以基于已积累的数据对药品的临床价值及其对专项基金预算的影响进行综合评估，与企业

[53] Flick, E. Dawn, Howard R. Terebelo, Susan Fish, Amani Kitali, Vrinda Mahajan, Melissa Nifenecker, Kristen Sullivan, Paul Thaler, Sarah Ussery, David L. Grinblatt. “The Value of Pharmaceutical Industry-Sponsored Patient Registries in Oncology Clinical Research.” *The Oncologist*, vol. 28, no. 8, 2023, pp. 657-663. https://doi.org/10.1093/oncolo/oyad110.

开展续约谈判，或者对已有仿制药或其它竞品上市的药品，通过国家医保谈判纳入基本医保药品目录管理。

（二）建议在国家统一指导下，优先选择罕见病诊疗防治体系较为完善的地区开展专项基金试点，为全国推广积累可复制、可推广的实践经验：

随着国家层面罕见病专项保障基金的顶层设计与筹备工作稳步推进，可在罕见病诊疗防治体系相对完善的地区率先开展专项基金试点。罕见病专项基金的建设可借鉴既往改革经验。例如在医保支付方式改革过程中，在国家级按疾病诊断相关分组（DRGs）付费系统开发的同时，各地同步积极探索，其积累的宝贵经验为随后全国统一技术规范的制定提供了坚实基础，并在试点阶段及时识别了潜在问题和风险因素。

基于此，在国家罕见病专项保障基金的构建过程中，地方同样可以同步启动试点工作，探索并试行国家专项基金方案，如专项基金规模测算、药品病例的真实世界数据收集、创新支付协议签署等。通过地方试点有助于发现实际运行中的问题并积累可推广的经验，从而有效控制专项基金在全国推开时可能面临的风险，提升国家方案设计的科学性与可行性，确保专项基金制度既契合中国国情，也能够真实反映中国罕见病诊疗的客观实践。

六、结语

完善罕见病医疗保障体系，是在新的发展阶段推进健康中国建设、落实全民健康目标的重要任务。中国已经构建了罕见病管理和诊疗能力提升的系统方案，通过一系列措施加快创新罕见病药物上市，并初步构建了多层次医疗保障格局；然而，现行体系罕见病高值药物支付保障能力方面仍存在明显

短板，高值罕见病药物保障不足已成为制约罕见病患者健康公平和医药创新良性循环的关键瓶颈。

在“十五五”新发展阶段，中国应采取“国家统筹设计、地方试点探索”相结合的模式建立罕见病专项基金，提升高值罕见病药物的保障水平，从而完善罕见病多层次医疗保障体系。在切实减轻罕见病患者疾病负担、防范因病致贫返贫的同时，为创新药企提供更为合理的商业回报，从而促进创新药产业发展，在“十五五”时期构筑产业发展新优势。

本报告提出的制度设计思路和政策建议，旨在为国家有关部门在“十五五”时期谋划和推进罕见病医疗保障体系完善提供决策参考。随着相关顶层设计的逐步落地和制度工具的不断完善，中国有望在罕见病领域创造罕见病多层次保障体系的“中国方案”，向国际社会展示制度优势，并为《WHA 决议》愿景的实现贡献“中国智慧”。

Improving the Medical Insurance System for Rare Diseases to Support the Achievement of the Strategic Goal of Health for All

AstraZeneca[1]

Executive Summary

The *Recommendations of the Central Committee of the Communist Party of China for Formulating the 15th Five-Year Plan for National Economic and Social Development*, adopted at the Fourth Plenary Session of the 20th Central Committee of the Communist Party of China (CPC), call for "advancing the 'Healthy China' initiative" as a strategic priority. General Secretary of the CPC Central Committee Xi Jinping emphasized that "people's health is a key indicator of China's progress towards building a modern socialist society."[2] The World Health Assembly (WHA) has recognized rare diseases as a global health priority.[3] The issue of rare diseases has therefore increasingly attracted widespread attention from the international community. The total population of rare disease patients in China is estimated to exceed 20 million.[4] Owing to the inherent complexity of rare diseases, difficulties in achieving timely and accurate diagnosis, and the scarcity of effective treatment options, patients generally experience significant health and economic burdens. Advancing the "Healthy China" initiative in the 15th Five-Year Plan period requires more equitable, accessible, high-quality, efficient, and affordable health and medical services for rare disease patients. At the same time, improving

[1] The views expressed in this report are those of the enterprise research and do not represent the official stance or opinions of the forum host and organiser.

[2] Xinhua. "Xi Jinping Stresses During Fujian Inspection the Need to Play a Greater Role in Serving and Integrating into the New Development Paradigm, and to Strive to Write a Fujian Chapter in the Comprehensive Building of a Modern Socialist Country". March 25, 2021. https://www.xinhuanet.com/politics/leaders/2021-03/25/c_1127254519.htm

[3] World Health Organization. "Rare diseases: a global health priority for equity and inclusion." May 27, 2025. https://apps.who.int/gb/ebwha/pdf_files/WHA78/A78_R11-ch.pdf.

[4] Frost & Sullivan, Illness Challenge Foundation. "2023 China rare disease industry trend observation report." 2023. https://www.frostchina.com/content/insight/detail/63f8f07f9c446c956d9754e3.

reimbursement capacity for high-cost rare disease drugs will not only address key bottlenecks in the development of innovative drugs, but also promote the growth of biomedicine as a strategic emerging industry, thereby building new competitive advantages for industrial development during the 15th Five-Year Plan period.

In recent years, through coordinated policy measures implemented by multiple central government ministries and agencies, China has made significant progress in rare disease prevention, treatment, and coverage. Such progress includes the establishment of a Multi-Tiered Medical Security System (MTMSS), in which Basic Medical Insurance (BMI) serves as the core pillar for insurance coverage, medical assistance functions as a safety net, and other mechanisms, including the supplementary medical insurance, Commercial Health Insurance (CHI), charitable donations, and medical mutual aid, are developed in a coordinated manner to provide additional coverage. However, at the current stage, the MTMSS still exhibits significant gaps for rare disease patients, underscoring the urgent need to establish a unified and systematic institutional framework at the national level to further enhance overall medical insurance coverage for patients with rare diseases. More specifically, the "basic coverage" mandate of the BMI limits its capacity to provide coverage for high-cost rare disease drugs, while regional disparities in benefit packages mean that some rare disease patients continue to face "last-mile" barriers to accessing needed medications. As for CHI, due to its profit-oriented risk management considerations, it is limited in its capacity to serve as a primary form of coverage for rare disease patients. Meanwhile, charitable donations, constrained by their limited funding and their operational models, struggle to provide stable and sustainable coverage.

A review of rare disease coverage systems across different countries suggests that the establishment of a dedicated rare disease fund to centrally manage payments for high-cost rare disease drugs is an extremely valuable model for China to learn from in its effort to optimize the MTMSS for rare disease patients and enhance payment capacity for high-cost rare disease drugs. In addition, as a result of sustained socioeconomic growth, China has reached a level of economic capacity that would

support the establishment of a dedicated fund for rare disease coverage. In recent years, several economically developed regions in China have already taken the lead in piloting dedicated rare disease funds, demonstrating the practical feasibility of this approach.

Drawing on international practices in rare disease coverage, the outcome of domestic pilot programs, and the existing foundation of China's MTMSS, this paper recommends that China progressively advance towards the establishment of a national dedicated fund for rare disease coverage. China could combine national-level coordinated design and regional pilot programs in its effort to establish a national dedicated fund for rare disease. While completing the top-level design and preparatory work for the dedicated fund at the national level, pilot programs may be launched in regions where rare disease prevention and treatment systems are relatively well developed. Regional pilot programs conducted under national-level unified guidance can lay a solid foundation for the nationwide rollout and coverage of a dedicated fund for rare disease coverage.

(I) Drawing on international practices in rare disease coverage, the outcome of domestic pilot programs, and the existing foundation of China's MTMSS, the design and implementation of a national dedicated fund for rare disease coverage could focus on the following aspects:

- The dedicated fund could clearly define the government departments responsible for nationwide fund oversight, management, and operation, while establishing systematic institutional arrangements across key steps in the process, including financing, access, payment, settlement, and evaluation, thereby ensuring a fund management model that is regulatable, sustainable, and traceable.
- The dedicated fund could establish stable and sustainable funding sources, for instance drawing from multiple channels, such as central fiscal allocations, public welfare lottery fund income, and donations from philanthropic organizations. The required funding scale may be calculated by referencing the number of diseases covered by the National Rare Disease Catalog, and the

number of patients registered in the National Rare Disease Direct Reporting System.

- At the national level, China could establish an expert committee to provide systematic review and evaluation for drugs intended to be covered by the dedicated fund, and to clearly define the drug categories and rare diseases that should receive priority coverage. To address the challenge posed by small patient populations in drug value assessment, authorities may consider adopting a more flexible framework when assessing the drugs intended to be covered by the dedicated fund.
- The dedicated fund could be allowed to enter innovative payment agreements with pharmaceutical companies and adopt financial outcome–based payment models to establish risk-sharing mechanisms for high-cost drugs, while setting a cap on individual out-of-pocket expenses. This mechanism not only keeps the patient financial burden within an affordable range but also strengthens the budget predictability and long-term sustainability of the fund.
- The dedicated fund could establish a mechanism to coordinate with the BMI, thus ensuring synergy and complementary coverage across the MTMSS. This would provide rare disease patients with continuous coverage throughout their entire life cycle. Real-world data collected during the implementation of innovative payment agreements can be used to assess the clinical value of rare disease drugs and their budgetary impact on the BMI fund, thereby providing evidence to support these drugs' participation in National Reimbursement Drug List (NRDL) negotiations.

(II) Under national unified guidance, China could select regions with relatively well-developed rare disease diagnosis, treatment, and prevention systems to pilot dedicated funds for rare disease coverage. Such pilots could provide replicable and scalable practical experience for the nationwide roll out of the dedicated fund.

Drawing on reform experience in the medical insurance sector, conducting local pilots in parallel with the development of a national policy framework can help identify potential challenges at an early stage and accumulate practical

implementation experience. Therefore, while the design for a national dedicated fund is still in development, dedicated fund pilots may be launched in regions with well-developed rare disease diagnostic and treatment capacity, covering fund size estimations, real-world data collection, and innovative payment agreement attempts. Regional dedicated fund pilots can help validate and strengthen the national-level fund design, ensuring that the national dedicated fund for rare disease is grounded in China's specific context and tailored to the realities of rare disease diagnosis and treatment in China.

I. Introduction

1. Healthcare Services for Rare Disease Patients Must Be Enhanced to Advance the "Healthy China" Initiative During The 15th Five-Year Plan Period

The *Recommendations of the Central Committee of the Communist Party of China for Formulating the 15th Five-Year Plan for National Economic and Social Development* (hereinafter referred to as *the Recommendations*), adopted at the Fourth Plenary Session of the 20th Central Committee of the CPC, call for "advancing the 'Healthy China' initiative" as a strategic priority. General Secretary of the CPC Central Committee Xi Jinping emphasized that "the people's health is a key indicator of China's progress towards building a modern socialist society."[5] Rare diseases represent a major public health challenge to human health. There are over 7,000 known rare diseases globally, of which approximately 80% are hereditary.[6] China has classified more than 1,400 conditions as rare diseases. While the prevalence of individual diseases is extremely low, due to China's large population, the total number of existing rare disease patients is estimated to exceed

[5] Xinhua. "Xi Jinping Stresses During Fujian Inspection the Need to Play a Greater Role in Serving and Integrating Into the New Development Paradigm, and to Strive to Write a Fujian Chapter in the Comprehensive Building of a Modern Socialist Country". March 25, 2021. https://www.xinhuanet.com/politics/leaders/2021-03/25/c_1127254519.htm

[6] The Lancet Global Health. "The Landscape for Rare Diseases in 2024." *The Lancet Global Health* 12, no. 3 (2024): e341. https://doi.org/10.1016/S2214-109X(24)00056-1.

20 million.[7] Moreover, rare disease patients universally bear heavy health and economic burdens due to the complexity of these diseases, the difficulty in their diagnosis, and the scarcity of their treatments. Rare diseases have a particularly prominent impact on children's health. Globally, 70% of rare diseases manifest in childhood.[8] In China, 36.07% of confirmed rare disease patients are under 18.[9] Improving the health of rare disease patients can contribute to improving children's health and enhancing the delivery of equitable and accessible health services to the entire population, thereby advancing the goals of the "Healthy China" initiative and fostering high-quality population development.

2. Improving Rare Disease Medical Insurance Coverage During the 15th Five-Year Plan Period Demonstrates the Unique Strength of Chinese Institutions in the Health Sector

In recent years, the international community has increasingly recognized the challenges that rare diseases pose to global health by incorporating rare diseases into the global health agenda. The 78th World Health Assembly (WHA), held in 2025, adopted the resolution, *Rare Diseases: A Global Health Priority for Equity and Inclusion,* (hereinafter referred to as the *WHA Resolution*) which for the first time recognized rare diseases as a global health priority,[10] The resolution recognizes "the importance of achieving universal health coverage, including for persons living with a rare disease". As one of the co-sponsors of this resolution, China has, in recent years, achieved remarkable results in rare disease prevention, treatment, and coverage through comprehensive, cross-departmental policy measures. With the *Recommendations* clearly setting the goal of "expanding high-level opening-up" and "providing more international public goods," China will further participate in and lead global health governance. China's efforts to optimize its medical insurance

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[7] Frost & Sullivan, Illness Challenge Foundation. "2023 China rare disease industry trend observation report." 2023. https://www.frostchina.com/content/insight/detail/63f8f07f9c446c956d9754e3.

[8] The Lancet Global Health. "The Landscape for Rare Diseases in 2024." *The Lancet Global Health* 12, no. 3 (2024): e341. https://doi.org/10.1016/S2214-109X(24)00056-1.

[9] Guo, Jian, Peng Liu, Limeng Chen, et al. "National Rare Diseases Registry System (NRDRS): China's First Nation-Wide Rare Diseases Demographic Analyses." *Orphanet Journal of Rare Diseases* 16, no. 1 (2021): 515. https://doi.org/10.1186/s13023-021-02130-7.

[10] World Health Organization. "Rare diseases: a global health priority for equity and inclusion." May 27, 2025. https://apps.who.int/gb/ebwha/pdf_files/WHA78/A78_R11-ch.pdf.

system for rare diseases not only actively reflects the priorities outlined in the *WHA Resolution*, but more importantly allows China to contribute its own "Chinese wisdom" for the advancement of a global rare disease response.

3. China's Rare Disease Prevention, Treatment, and Coverage Efforts Have Achieved Remarkable Results

China has made significant progress in rare disease prevention, treatment, and coverage, implementing a series of policy measures across the medical service, medical insurance, and pharmaceutical sectors. These measures have comprehensively enhanced China's capacity for diagnosing, treating, and covering for rare diseases. In the pharmaceutical sector, the National Medical Products Administration (NMPA) has optimized the review and approval processes to incentivize innovative rare disease drug development. The NMPA has also established expedited import channels for clinically urgently needed drugs, accelerating the introduction of rare disease drugs that are under development or already on the market in other countries or regions. In terms of diagnosis and treatment management, the National Health Commission (NHC) has carried out systematic initiatives, such as publishing the National Rare Disease Catalog and formulating standardized diagnostic and treatment guidelines. The NHC has also organized a national collaboration network for the diagnosis and treatment of rare diseases and built data platforms. These efforts represent a comprehensive effort to improve care for rare disease patients spanning diagnosis, medical treatment quality control, insurance coverage, disease management, and research. In the medical insurance sector, the National Healthcare Security Administration (NHSA) has established a three-tiered medical insurance system composed of BMI, serious disease insurance, and medical aid. China has enhanced its ability to provide insurance coverage for rare disease patients by incorporating rare disease drugs into the NRDL and exploring the creation of the Commercial Health Insurance Innovative Drug List (CHIIDL).

II. Optimizing the Rare Disease Medical Insurance System Will Have a Critical Impact on the Innovative Development of China's Biomedical Industry in the 15th Five-Year Plan Period

Upgrading rare disease diagnosis and treatment capacity goes hand in hand with pharmaceutical innovation. Given that approximately 95% of rare diseases still lack effective treatment options, rare diseases have become one of the global focal points for new drug research and development (R&D).[11] Statistics from Market Monitor projected that the global orphan drug market would reach RMB 1.55 trillion in 2025.[12] Research into the pathogenesis of rare diseases and innovative therapies can further drive technological innovation in the treatment of common diseases, offering great scientific and economic value. For example, the discovery of the novel lipid-lowering drug class, PCSK9 inhibitors, originated from studies of rare genetic mutations.[13]

Rare diseases represent a high-potential field for the innovative development of China's biopharmaceutical industry. China's large population, rapid patient recruitment, low clinical research costs, and swift advances in artificial intelligence provide unique advantages in rare disease drug R&D that could allow China to achieve "leapfrog" advancement in innovative drug development for rare diseases. China has made significant progress in rare disease drug innovation, supported by policies that stimulate R&D and strengthen the entire innovation value chain. These advancements in R&D, combined with the clinical value of these drugs, have earned international recognition. Of the 481 drugs that received Orphan Drug Designation from the U.S. FDA in 2024, 68 were submitted by Chinese pharmaceutical companies.[14] From the perspective of countries of origin for innovative drugs,

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[11] Zhang Shuyang, Zhang Xue. "Recent Polices and Practice in Rare Diseases in China." *Journal of Rare Diseases*, 2022, 1(1): 1-6. https://doi.org/10.12376/j.issn.2097-0501.2022.01.001.

[12] Global Market Monitor. "Report on 2025 Orphan Drug Market Development Trends: Market Size, Pricing Trends, and Competitive Analysis." August 28, 2025. https://www.sohu.com/a/929318840_122448164.

[13] Feng Siqin, Wang Yifei, Chen Peipei, Zhang Zhiyu, and Zhang Shuyang. "Research Progress on the Pleiotropic Effects of PCSK9 Inhibitors." *Chinese Journal of Cardiology*, Vol. 50, No. 3, 2022. https://csc.cma.org.cn/art/2022/7/6/art_620_45976.html.

[14] DrugTimes. "481 Drugs! A Surge in FDA Orphan Drug Designations in 2024, With China Ranking Second." March 25, 2025. https://pharm.jgvogel.cn/c1498564.shtml.

China ranks second. This underscores the country's emergence as one of the world's leading forces in biopharmaceutical innovation.

For the time being, the development of innovative drugs in China still faces several challenges, such as the discrepancy between the pricing expectations of innovative pharmaceutical companies and the reimbursement capacity of medical insurance, as well as the lack of diversified reimbursement channels.[15] Pricing and reimbursement policies have become major factors constraining the R&D, commercialization, and international market expansion of indigenous rare disease drug innovations. The *Recommendations* state that China will "comprehensively enhance indigenous innovation capacity and continuously foster new quality productive forces." They also call for "accelerating the construction of a 'Healthy China' in the 15th Five-year Plan period," and identified "supporting the development of innovative drugs and medical devices" as one of the key measures to achieve this goal. Therefore, enhancing the rare disease medical insurance system is crucial to optimizing the innovative drug ecosystem, advancing the biomedical industry as a strategic emerging industry, and shaping new industrial advantages during the 15th Five-Year Plan period.

This report, based on the current condition of China's rare disease medical insurance system and the challenges it currently faces, systematically reviews international and domestic practice. The report proposes policy recommendations for establishing a national dedicated fund for rare disease coverage. It aims to serve as a reference to support policymaking for the establishment of a rare disease medical insurance system that is more equitable, efficient, and sustainable during the 15th Five-Year Plan period. At the same time, it also seeks to support pharmaceutical innovation and the achievement of China's goal of health for all.

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[15] National Healthcare Security Administration. "Transcript of the Press Conference on *Several Measures to Support the High-Quality Development of Innovative Drugs*." July 1, 2025. https://www.nhsa.gov.cn/art/2025/7/1/art_14_17065.html.

III. Progress and Challenges in the Construction of a Medical Insurance System for Rare Diseases in China

(I) Progress in Building a Medical Security System for Rare Diseases: Initial Establishment of a Multi-Tiered Security Framework

China has achieved remarkable accomplishments in the prevention, treatment, and coverage of rare diseases, including the establishment of an MTMSS for rare disease centered on BMI, underpinned by medical aid, and supplemented by serious disease insurance, CHI, charitable donations, and medical mutual aid.

1. Inclusion of Rare Disease Drugs in the NRDL through Negotiation

Since 2017, the NHSA and its predecessor have incorporated rare disease drugs into the NRDL through price-negotiations, effectively enhancing coverage for rare disease treatments. By the end of 2024, a total of 126 rare disease drugs covering 68 types of rare diseases had been included in the NRDL, accounting for roughly two thirds of all rare disease drugs approved in the Chinese market.[16] The 2025 edition of the NRDL added another 10 rare disease drugs, further expanding coverage.[17]

2. Local Initiatives: Providing Supplementary Coverage for Drugs Not Included in the NRDL through Serious Disease Insurance and Dedicated Funds

Local governments have actively explored supplementary coverage for rare diseases that have yet to be included in the NRDL, such as establishing dedicated funds and expanding the scope of serious disease insurance. These efforts have, to some extent, alleviated the disease burden for certain rare disease patients. For example, Zhejiang Province's dedicated fund for rare diseases provides coverage for high-cost, innovative rare disease drugs not yet included in the NRDL, covering patients with Gaucher disease, Pompe disease, Fabry disease, and phenylketonuria. Hunan Province incorporated two drugs for the treatment of Gaucher disease and

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[16] Frost & Sullivan, Illness Challenge Foundation. "Observation Report on the Trends of China's Rare Diseases." February 28, 2025. https://www.frostchina.com/content/insight/detail/67bed1ec7ed30cc08c184b97.

[17] Economic Daily. "Medical Insurance and Commercial Insurance 'Dual Drug Lists' Strongly Support Innovation—High-Quality and New Drugs Reach the Public Faster." January 3, 2026. https://www.news.cn/fortune/20260103/a458cb7c27b3499c823fc43b29777a3e/c.html.

Pompe disease, both not yet included in the NRDL, into the reimbursement scope of local serious disease insurance schemes. Through negotiations and cost-sharing arrangements with pharmaceutical companies, serious disease insurance covers 50% of the drug costs, companies cover about 40%, and individual out-of-pocket expenses are limited to RMB 30,000.

3. City Supplemental Insurance: Provides Supplementary Coverage for Rare Disease Patients

Inclusive supplementary CHI products that are underwritten by insurance companies and led by local governments provide additional coverage for rare disease patients. By July 31, 2025, a total of 313 City Supplemental Insurance (CSI) products had been launched, of which 202 are still in operation.[18] Over the past three years, more than 50% of these CSI products have included rare disease drugs.[19] Most CSI products either reimburse out-of-pocket expenses for rare disease drugs included in the NRDL or cover high-cost innovative rare disease drugs not included in the NRDL.[20]

In December 2025, the NHSA released the first edition of the M, which included 19 drugs. Six of these drugs are indicated for the treatment of rare diseases such as neuroblastoma, Gaucher disease, and phenylketonuria. This initiative has laid the foundation for the differentiation of CHI and BMI, further optimizing the MTMSS.[21]

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[18] National Business Daily. "Prices Rise, Products Shrink, but the "Core Base" Holds Steady: Inclusive Health Insurance Enters a 'Mature Phase'." October 22, 2025. https://www.nbd.com.cn/articles/2025-10-22/4101569.html.

[19] Health News. "How Can Coverage for Rare Disease Medicines Go Further? — A Conversation with Mao Ningying, Associate Dean of School of International Pharmaceutical Business at China Pharmaceutical University." November 14, 2025. https://www.jkb.com.cn/horizon/2025/1114/507068.html

[20] Health News. "How Can Coverage for Rare Disease Medicines Go Further." November 14, 2025. https://www.jkb.com.cn/horizon/2025/1114/507068.html.

[21] Yicai. "Six Rare Disease Drugs Included in the First Commercial Insurance Innovative Drug List: How Can More Commercial Insurers Be Attracted to Participate?" December 10, 2025. https://news.qq.com/rain/a/20251210A05VUO00.

4. Medical Aid and Charitable Efforts: Providing a Safety Net for Rare Disease Patients

Local governments provide dedicated subsidies for rare disease patients through medical assistance programs, building a safety net against catastrophic health expenditures and reducing the risk of poverty caused by illness. For example, Foshan Municipality provides reimbursement for over 50 drugs and therapeutic foods for rare diseases, including Gaucher disease, Fabry disease, and type II glycogen storage disease, through medical aid programs. After receiving reimbursement from BMI, serious disease insurance, and supplementary medical insurance programs, eligible beneficiaries will pay any remaining expenses out-of-pocket. Following this, such patients may receive medical assistance from medical security authorities in their district of household registration for 80% of the costs they incurred, with an annual cap of RMB 300,000.

Philanthropic organizations and public welfare initiatives also play vital roles in providing personalized and diversified support for rare disease patients. In 2023, the Foshan Medical Security Association, Foshan Maternal and Child Health Hospital, and the Illness Challenge Foundation (ICF) jointly launched a pilot project for charitable medical aid and comprehensive services for rare diseases, under the guidance of the Foshan Municipal Healthcare Security Bureau. This project created the first municipal-level model integrating medical insurance, charitable medical aid, and comprehensive services for rare disease patients. The ICF stationed medical social workers specializing in rare diseases at Foshan Maternal and Child Health Hospital to provide services such as patient screening, assistance applications, medication support, and psychological counseling. The ICF also worked to embed Foshan's rare disease medical aid process into healthcare institutions' medical service and social work procedures. Further, the foundation regularly assists the Foshan Medical Security Association in verifying patients' medical insurance reimbursement data to avoid duplicate funding.

(II) Major Challenges and Limitations in China's Medical Insurance System for Rare Diseases

Although China has established an MTMSS for rare diseases, there is significant room for improvement in overall coverage capacity, particularly in the reimbursement mechanisms for high-cost rare disease drugs. Research shows that approximately 1.1% of rare disease patients in China face extremely high disease burdens,[22] with out-of-pocket direct expenses amounting up to 2460.81% of their average annual household income.[23] As for the 70.2% of rare disease patients facing high disease burdens,[24] their out-of-pocket direct medical expenses on average amount to 95.88% of their annual household income.[25] Considering that approximately 60% of rare disease patients' households have an annual income of less than RMB 30,000,[26] the "catastrophic-level household health expenditures" caused by rare diseases may drive families into or back into poverty. Consequently, the creation of a dedicated payment mechanism for high-cost rare disease drugs constitutes an urgent priority for China's rare disease patient community.

1. BMI's Mandate to Provide Basic Coverage Limits Its Ability to Cover High-Cost Drugs

Rare disease drugs are costly and time-consuming to develop. However, the small number of patients causes the price of rare disease drugs to be generally high. At present, 65 drugs treating 58 rare diseases have yet to be covered by BMI. Among them are 19 drugs, covering 26 rare diseases, for which none of the available

[22] Yu J, Chen S, Zhang H, et al. "Patterns of the health and economic burden of 33 rare diseases in China: nationwide web-based study." *JMIR public health and surveillance*, 2024, 10: e57353.
[23] Patients with rare diseases who bear an extremely high disease burden are characterized by a significantly higher likelihood of reporting moderate to severe physical and mental health problems, alongside markedly elevated average out-of-pocket medical expenditures—relative to household income—stemming from direct medical costs, direct nonmedical costs, and indirect costs.
[24] Patients with rare disease who bear high disease burden are primarily characterized by a higher probability of reporting moderate to severe health problems, while disease-related average economic expenditures are also relatively high when measured against household income.
[25] Yu J, Chen S, Zhang H, et al. "Patterns of the health and economic burden of 33 rare diseases in China: nationwide web-based study."
[26] China National Radio. "Two Sessions 2025 | NPC Deputy Wu Huangan: Establishing a Multitiered Security System for Rare Diseases." March 10, 2025. https://roll.sohu.com/a/869005026_362042.

treatments are covered by BMI and which carry particularly high costs.[27] The vast majority of rare disease patients whose treatments are not covered by BMI either forgo treatment entirely or resort to inadequate dosing or incomplete treatment. These practices severely compromise patients' health and survival.[28]

2. The "Last Mile" Challenge: Regional Disparities in BMI Benefits Leave Some Rare Disease Patients Facing Significant Out-of-Pocket Burdens

Although some rare disease drugs have been included in the NRDL, disparities in BMI benefits across regions mean that some patients still face "last mile" barriers in accessing these drugs. Many rare diseases require long-term treatment and follow-up care in outpatient settings. Nonetheless, a study examining the inclusion of chronic and special diseases in outpatient medical insurance across 300 prefecture-level cities in China shows that only 24 rare diseases are covered under local outpatient medical insurance policies.[29] In most cities, fewer than four rare diseases are covered. As a result, many rare disease patients are still unable to receive treatment or benefit from reimbursement in outpatient settings. Additionally, significant regional discrepancies persist in outpatient reimbursement rates for rare disease drugs included in the NRDL. Outpatient reimbursement rates under the Urban and Rural Resident Basic Medical Insurance (URRBMI) are relatively low. In some provinces and cities, the reimbursement policies for outpatient medical insurance and serious disease insurance are not effectively integrated, resulting in significant out-of-pocket expenses for rare disease patients.

3. CHI Has Limited Capacity to Serve as an Effective Supplementary Mechanism for Rare Disease Coverage

Currently, CHI coverage in China is relatively low, reaching only 0.7% of GDP, far below the level observed in developed economies.[30] As a profit-driven business,

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[27] Frost & Sullivan, Illness Challenge Foundation. "Observation Report on the Trends of China's Rare Diseases."
[28] Ibid.
[29] Xu, Juan, Mingren Yu, Zhiguo Zhang, Shiwei Gong, Bingqin Li. "Is Sub-National Healthcare Social Protection Sufficient for Protecting Rare Disease Patients? The Case of China." Front. Public Health, 2023;11.
[30] Caizhongshe. "Guolian Securities: China's Effective Coverage Rate for Commercial Health Insurance Remains Relatively Low." January 17, 2025. https://news.qq.com/rain/a/20250117A029QM00.

CHI operates by the law of large numbers to disperse low-probability individual health risks through expanding the insured pool.[31] Approximately 80% of rare diseases are hereditary, thus the proportion of high-risk individuals within the rare disease population is significantly higher. To avoid the risk of "adverse selection," in which healthy individuals have weak incentives to enroll while high-risk patients with preexisting conditions are more likely to participate, commercial health insurers often manage risks by imposing measures such as exclusions for preexisting conditions. As a result, CHI is unable to provide effective supplementary coverage for patients with rare diseases.

CSI led by local governments are currently undergoing market adjustment which includes the optimization and evolution of product design. The coverage and benefit offerings across different regional CSI products vary greatly, resulting in different levels of coverage offered to patients living with rare diseases. By July 2025, 47 CSI products included coverage for rare diseases, accounting for 25% of all CSI products.[32] CSI products in 42 cities provided coverage for rare diseases, representing 30% of all cities offering such products. For patients living with rare diseases, CSI products cannot yet serve as a stable and reliable supplementary means of insurance coverage. For this reason, some rare disease patients still face heavy financial burdens.

4. Charitable Donations Cannot Serve as the Primary Source of Coverage for Rare Disease Patients

Charitable efforts play an important role in bridging the "last mile" of rare disease drug coverage, but due to their inherent limitations, they cannot serve as a stable or sustainable main source of coverage. First, the source of funds is unstable. Over 60% of charitable funds rely on corporate donations, which are highly influenced by

[31] "Current Status, Challenges, and Policy Responses of Commercial Health Insurance in China's Rare Disease Medical Coverage." *Journal of Rare Diseases*. January 14, 2022. https://jrd.chard.org.cn/cn/article/doi/10.12376/j.issn.2097-0501.2022.01.014.

[32] Heguan Yiliao. "Commercial Insurance Innovative Drug Applications: Candidate List for Rare Disease Therapies and the Current Distribution of Access Under Inclusive Health Insurance Programs." July 2, 2025. https://mp.weixin.qq.com/s/L8T1K5ILOUbDRA4NrQ9ODg.

public attention and economic conditions.[33] Further, only about 18% of donations are directed towards healthcare. Surveys indicate that public awareness of rare diseases is very limited, with approximately 75% of the public indicating awareness of only three or fewer rare diseases.[34] This significantly restricts the ability of charitable donations to effectively support rare disease patients. Second, coverage is fragmented. Charitable programs often target specific diseases, with inconsistent scopes of coverage and application standards. Public welfare projects primarily focus on major diseases such as malignant tumors, with only 5% of donations allocated to rare diseases.[35] Third, rare diseases often require long-term or even lifelong treatment, but most charitable programs provide funding for only one year while also often imposing funding caps. This short-term, fixed-amount funding model cannot meet rare disease patients' inelastic demand for long-term medication. As a result, charitable efforts primarily play a role in filling gaps and addressing urgent needs within the rare disease coverage system, rather than assuming systemic payment responsibilities.

In summary, significant gaps remain in China's MTMSS in providing insurance coverage to rare disease patients. The levels of coverage accessible to rare disease patients also vary greatly among regions. Therefore, it is imperative to establish a unified, sustainable reimbursement mechanism at the national level to systematically enhance medical insurance coverage for rare disease patients in China, while also providing sustained momentum for the development of innovative drugs.

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[33] Song Zonghe. "China Charitable Donations Report, 2019–2020," in *Charity Blue Book: China Charity Development Report 2021*, edited by Yang Tuan and Zhu Jiangang. *Social Sciences Academic Press*, December 2021.

[34] China Global Philanthropy Institute. "Public Awareness Survey Report on Rare Diseases." February 28, 2023. https://www.cgpi.org.cn/index/hotnewstext/id/1335.html

[35] Deloitte. Patient Assistance Joint Action Alliance. *Yearbook of patient assistance in China 2021*.

IV. International and Domestic Policy Best Practices

Globally, rare disease coverage systems generally fall into three main models: dedicated fund–based multi-payer schemes, universal health insurance–based coverage, and commercial insurance–based coverage.

- France has adopted a coverage model that relies on statutory health insurance funds and has developed a list system for long-term diseases (Affections de Longue Duree, ALD). Within this system, 100% of expenses related to diseases included in the ALD 30 list are reimbursed.[36] The ALD 30 list contains several types of rare diseases. For diseases not included in the ALD 30 that require more than six months of costly medical treatment, patients can apply for 100% of expenses to be reimbursed through health insurance channels on a case-by-case basis.
- In the United States, most of the population is covered by commercial insurance. Low-income and elderly groups are often covered by Medicaid and Medicare, respectively. The *Affordable Care Act* stipulates that most commercial health insurance plans cannot deny coverage based on pre-existing conditions, including rare diseases.[37] However, the Act does not specify which rare disease treatments must be reimbursed by commercial health insurance programs, nor does it specify reimbursement standards. For elderly patients covered by Medicare, individuals that reach the "catastrophic expenditure threshold" (USD 2,100 in 2026) can receive 95% reimbursement for costly prescription drugs.[38] For Medicaid, each U.S. state independently manages its own drug reimbursement policy and list, resulting in significant state-to-state differences in the coverage of orphan drugs for low-income patients.[39]

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[36] Directorate of Legal and Administrative Information (Prime Minister). "Management of a long-term illness (ALD) by the Health Insurance." Accessed March 10, 2026. https://www.service-public.gouv.fr/particuliers/vosdroits/F34068?lang=en.
[37] U.S. Department of Health and Human Services. "Pre-Existing Conditions." March 17, 2022. https://www.hhs.gov/healthcare/about-the-aca/pre-existing-conditions/index.html.
[38] Wreschnig, Laura A. "Medicare Part D Prescription Drug Benefit." *Congressional Research Service.* November 14, 2023. https://www.congress.gov/crs-product/R40611.
[39] KFF. "State Medicaid Drug Review Responsibilities." July 1, 2019. https://www.kff.org/state-health-policy-data/state-indicator/medicaid-drug-review-responsibilities/.

- Italy established a dedicated fund for rare diseases to supplement its universal health insurance program, elevating the country's ability to provide coverage for rare disease medical expense.[40] Italy's dedicated fund for rare diseases utilizes a portion of the drug promotion taxes levied on pharmaceutical companies and grants from the National Health Fund to reimburse up to 100% of expenses for rare disease drugs.

Based on common international models of rare disease medical insurance coverage and China's existing MTMSS, the model of a rare disease dedicated fund may serve as a valuable reference for further optimizing China's MTMSS for rare diseases.

(I) An examination of dedicated funds for rare diseases in multiple countries reveals several common features in their operating models and management structures:

1. Legal Basis and Stable Funding Sources for the Establishment of Rare Disease Funds

A dedicated fund for rare diseases requires earmarked and sustainable financing sources, typically derived from national tax revenues and/or transfers from universal health insurance funds. For this reason, many countries have enacted laws and regulations related to rare diseases to provide the legal basis and institutional safeguards for the establishment and operation of such funds. For example, in 2024 Egypt amended legislation that manages the Emergency Medical Fund, providing an institutionalized fiscal guarantee for rare disease treatment.[41] This legislation stipulates that the Fund must cover the costs of specific rare disease drugs and treatments. Under this framework, treatment for rare disease patients no longer depends on fragmented programs, charitable organizations, or temporary funding, thus creating a long-term and sustainable coverage mechanism.

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[40] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. "The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy." *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

[41] Tinnion, Violet. "Egypt Expands Rare Disease Funding through Emergency Medical Fund." FrontierView, January 24, 2024. https://app.frontierview.com/insightBite/1881/egypt-expands-rare-disease-funding-through-emergency-medical-fund.

2. Diseases Covered by the Dedicated Fund Are Determined Based on Established Rare Disease Definitions or Officially Recognized Rare Disease Catalogs

There is no single unified and universally accepted definition of "rare diseases" at the global level. Many countries have either defined rare diseases in relevant legislation and national rare disease action plans or established national rare disease catalogs. Such legal definitions or catalogs serve as the primary basis for determining which rare diseases are eligible for coverage under dedicated national funds, universal health insurance schemes, or commercialized medical insurance. For example, Russia defines rare diseases as those with a maximum prevalence of no more than 10 cases for every 100,000 people. The Ministry of Health of the Russian Federation also formulated a rare disease catalog, which currently includes approximately 290 diseases. Building on this framework, Russia's "Circle of Kindness" Foundation has formulated a separate catalog specifically for the country's dedicated fund for rare disease patients under the age of 19, which the Foundation administers. This catalog is revised at least once every two years. From 2021 to 2025, the number of diseases included in this catalog increased from 44 to 101, and the fund has provided medication to more than 29,000 children.

3. Covering Drugs that have Received Approval in Other Countries and Drugs that Have Received Domestic Market Approval to Encourage Innovation

To ensure that rare disease patients can access innovative therapies in a timely manner, dedicated national rare disease funds in many countries cover drugs that have not yet been approved in their domestic markets, in addition to those that have already been approved. This gives patients timely access to the latest treatment options for rare diseases. Italy provides an illustrative example of this practice.[42] Italy's dedicated fund for rare diseases not only covers drugs approved by the European Medicines Agency (EMA) that have not yet been introduced in the Italian market, but also covers drugs already approved in the Italian market. More than 80% of the orphan drugs approved by the EMA can be reimbursed in Italy. While Italy's

[42] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. "The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy." *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

spending on orphan drugs is comparable to the European average, drug prices are relatively lower in Italy.

For orphan drugs approved by the EMA but not yet marketed in Italy, the country established the "AIFA 5% Fund," reimbursing 100% of actual expenses incurred by patients. The fund is financed through a 5% tax on pharmaceutical companies' promotional expenditure. Of this tax revenue, 50% is allocated to the "AIFA 5% Fund."[43] Pharmaceutical companies do not need to conduct pricing negotiations to access the "AIFA 5% Fund." Rather, only hospitals and healthcare institutions need to submit applications for drugs to be reimbursed.

If the Italian Medicines Agency (Agenzia Italiana del Farmaco, AIFA) determines a product that has already been introduced to the Italian market is "fully innovative," Italy's Fund for Innovative Medicines reimburses 100% of costs incurred within the first 36 months after the product's market approval.[44] Approximately one-third of the innovative medicines recognized by AIFA each year are orphan drugs. The Fund is financed by an annual allocation of EUR 1.3 billion (~RMB 10.7 billion) from the National Health Fund.[45]

4. Establishing Risk-Sharing, Innovative Payment Agreements with Pharmaceutical Companies to Accelerate Access to Innovative Therapies While Maintaining Fund Sustainability

Dedicated funds for rare diseases in other countries and regions use budget management and risk-sharing agreements to help health insurance authorities manage the risk of overdrawing the dedicated fund. At the same time, they offer coverage to patients who are the most in need of treatment but are unable to afford it. These measures significantly reduce the burden of medical expenses on families

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[43] Since the promulgation of *Decree No. 175/2021*, the tax rate has been raised to 7%, with the additional 2 percentage points earmarked to support public research related to rare diseases, neglected diseases, and advanced therapies.

[44] Vogler, Sabine. "Payer Policies to Support Innovation and Access to Medicines in the WHO European Region." Copenhagen Ø, Denmark: World Health Organization. 2022. 4. Case studies. https://www.ncbi.nlm.nih.gov/books/NBK587872/.

[45] Prior to 2022, the Fund for Innovative Medicines consisted of two separate entities for oncology and non-oncology drugs. In 2022, these two funds were merged into one single fund encompassing all innovative medicines, with a total budget of EUR 1 billion for that year.

of rare disease patients. The risk-sharing model is an innovative payment mechanism widely used internationally for rare disease and oncology drugs. This model not only accelerates clinical application of new drugs but also improves the efficiency of health insurance funds and mitigates financial risk. For example, in September 2018, the health insurance administration of Taiwan, China added managed entry agreements (MEAs) into its *Regional Health Insurance Drug Reimbursement Items and Standards* as an "other agreement mechanism."[46] The MEAs can be signed either based on clinical or financial outcomes. The maximum term for these agreements is five years, with the option to renew upon expiration. Companies are allowed to propose agreement terms and negotiate specific conditions with the local health insurance administration. Any drug costs that companies are required to reimburse under these agreements are incorporated into the settlement of patients' total medical expenses.

5. Establishing Mechanisms for Coordination with Existing National Health Insurance Schemes to Provide Patients with Continuous Coverage, While Ensuring that Patients Do Not Need to Pay Out-of-Pocket or That Their Out-of-Pocket Expenses Remain Within an Affordable Range

Developed countries and regions that provide rare disease coverage primarily through dedicated funds or universal health insurance typically reimburse 100% of drug expenses for patients with rare diseases. Notable examples of countries implementing such practices include France[47] and Italy[48]. In some countries and regions, dedicated funds are responsible for covering the costs of rare disease drugs during specific periods or stages. By establishing mechanisms to coordinate the dedicated fund with national health insurance scheme, these countries and regions were able to provide rare disease patients with continuous coverage, effectively

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[46] WANG Meifeng, WANG Haiyin, CONG Lixuan, XIE Chunyan, LIU Xin, JIN Chunlin. Study on the experience and enlightenment of medical insurance payment of innovative drugs in Taiwan, China. *World Clinical Drug*. 2021, 42(03): 223-228 https://doi.org/10.13683/j.wph.2021.03.014

[47] Service Public, Management of a long-term illness (ALD) by the Health Insurance, Accessed on March 5, 2026 https://www.service-public.gouv.fr/particuliers/vosdroits/F34068?lang=en

[48] People with rare or chronic diseases, including HIV, and pregnant women are exempt from cost-sharing for treatments related to their condition. The Commonwealth Fund "International Health Care System Profiles: Italy". Accessed on March 14, 2026. https://www.commonwealthfund.org/international-health-policy-center/countries/italy

reducing their out-of-pocket medical expenses. For example, in Italy, as noted above, 100% of patients' expenses related to drugs designated by the AIFA as "fully innovative" are reimbursed by the Fund for Innovative Medicines for the first 36 months after their market approval. After this period, each region will provide reimbursement for these drugs according to its local medical insurance budget at prices agreed upon between the AIFA and pharmaceutical companies.

In the early months of new drugs' entry into the Italian market, Italy systematically collects and evaluates real-world data on their usage through MEAs and the corresponding patient case registries. The collected data provides critical evidence for future negotiations on drug pricing and for the determination of reimbursement standards. Most medicines approved for reimbursement by AIFA are accompanied by MEAs. To support the implementation of MEAs, Italy established a national patient case registry in 2005 to document cases involving drugs under agreement. By the end of 2019, the registry had recorded 283 cases categorized by indication, covering 159 drugs. Of these entries, 88 addressed rare diseases, involving 52 orphan drugs.[49] Based on information collected in this registry, health insurance authorities can provide reimbursement based on the drugs' clinical outcomes or require companies to refund medicine costs. In the future, aggregated analyses of this data can further support the selection of optimal treatment regimens.

(II) Domestic Pilot Programs Prove the Feasibility of the Dedicated Fund Model

In recent years, economically developed regions have taken the lead in exploring mechanisms for rare disease coverage. Among them, Zhejiang Province adopted a dedicated fund model featuring provincial-level coordination with independent funding sources. Zhejiang's provincial fund has demonstrated strong financing capacity and a relatively high level of coverage, while maintaining stable operations

[49] Xoxi, Entela, Karen M. Facey, and Americo Cicchetti. "The Evolution of AIFA Registries to Support Managed Entry Agreements for Orphan Medicinal Products in Italy." *Front. Pharmacol*., vol. 12, 2021. https://doi.org/10.3389/fphar.2021.699466.

and balanced expenditures. This fund provides the following best practices that may be instructive for establishing a national dedicated fund for rare diseases:

- **Earmark funding to ensure a stable and sustainable source of funds:** Each year, a one-off transfer of RMB 2 per person is made from serious disease insurance funds to the provincial rare disease fund. The fund is managed under a sub-account within the dedicated fiscal account of the Zhejiang provincial medical insurance fund, allowing for separate management and independent accounting.
- **Define the covered diseases (Gaucher disease, Pompe disease, Fabry disease, and phenylketonuria) through expert appraisal:** The dedicated fund's expert committee is composed of rare disease clinicians, scholars, and medical insurance policy experts.
- **Establish tiered reimbursement rates and out-of-pocket limits for individuals:** Drug expenditures incurred by insured individuals within a settlement year are reimbursed on an accumulated, tiered basis. The reimbursement rate is 80% for expenses up to RMB 300,000, 90% for expenses between RMB 300,000 and RMB 700,000, and fully covered expenses exceeding RMB 700,000. The individual out-of-pocket expense limit is capped at RMB 100,000.
- **Coordination with medical aid:** After receiving reimbursement from the dedicated rare disease fund, eligible patients may apply for any remaining expenses to be covered through medical aid by the local government at their place of medical insurance enrollment.
- **Establish a risk-sharing mechanism with pharmaceutical companies:** Zhejiang conducts negotiations for rare disease drugs with pharmaceutical companies, while also setting an annual ceiling for the total reimbursed expenditure for high-cost rare disease drugs. Since its establishment, Zhejiang's dedicated rare disease fund has operated in a stable manner, maintaining an annual surplus from 2020 to 2024.

V. Policy Recommendations

As a result of sustained socioeconomic growth, China has reached a level of economic capacity that would support the establishment of a dedicated fund for rare disease coverage. When Egypt revised its medical emergency fund legislation in 2024 to provide an institutionalized fiscal guarantee for the treatment of patients with rare diseases, its GDP per capita was USD 3,338. When Russia established the "Circle of Kindness" Foundation in 2021 to finance the treatment costs of rare disease patients under the age of 19, its GDP per capita was USD 12,425.[50] During the 14th Five-Year Plan period, China's GDP per capita increased from USD 10,000 to more than USD 13,000,[51] placing China among the ranks of upper-middle-income countries.[52] Therefore, China should consider exploring the introduction of a dedicated fund at the national level for rare diseases to further optimize the "1+3+N" MTMSS. This would strengthen the payment capacity of the "N" components (i.e., other complementary coverage mechanisms) for high-cost rare disease drugs, thereby providing rare disease patients with accessible and affordable healthcare services.

It is recommended that China adopt an approach combining national-level design with local pilot implementation. Top-level design and preparatory work for a national dedicated fund for rare diseases can be completed at the national level. In the meantime, governments in regions with relatively well-developed rare disease diagnosis, prevention and treatment systems could conduct pilots on the dedicated fund model. Regional pilots could conduct trials on key elements and key processes of a dedicated fund, such as funding scale estimation, innovative payment mechanism arrangements, and the establishment of patient case databases. Through these pilots, replicable and scalable experiences can be accumulated. Under unified

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[50] World Bank Group. "World Bank Open Data: GDP per capita (current US$) - Russian Federation, Egypt, Arab Rep." 2024. https://data.worldbank.org/indicator/NY.GDP.PCAP.CD?locations=RU-EG.

[51] Xinhua. "Press conference on economy held during 4th session of 14th NPC." March 6, 2026. https://www.news.cn/politics/20260306/81996586d4ac4e4faf95d0431089db68/c.html.

[52] China Economic Net. "National Bureau of Statistics: 'China's Per Capita GDP Ranks Among the Leading Upper-Middle-Income Countries'." October 1, 2025. http://www.ce.cn/xwzx/gnsz/gdxw/202510/t20251001_2501161.shtml.

national guidance, the advantages of local experimentation can be fully leveraged to lay a solid foundation for the nationwide rollout and coverage of a dedicated fund for rare disease coverage.

(I) Building on prevailing international practices, the outcomes of local pilot programs in China, and the practical foundations of China's MTMSS for rare diseases, the design and implementation of the dedicated fund may focus on the following aspects:

Improving Top-Level Design and Establishing an End-to-End Management Mechanism: The management of the fund should adopt a cross-departmental coordination model, with dedicated national-level bodies responsible for oversight, management, and implementation. Responsibilities for other relevant departments would be clearly defined. National-level departments would be responsible for making systematic institutional arrangements regarding financing mechanisms, covered populations, eligibility criteria, benefit levels, information platforms, administrative rules, and complementary measures. The operation of the fund must be regulatable, sustainable, and traceable. The fund would need to establish an end-to-end management mechanism covering key financial processes such as fundraising, access, payment, claims settlement, and evaluation. Operating the fund according to these principles would ensure that workflows and personnel are subject to oversight, financing sources and scale are sustainable, and the use of funds and patient information are traceable.

Establishing Stable and Sustainable Financing Sources: China's BMI fund faces ongoing pressure in balancing its revenue and expenditures. CHI, according to its inherent business logic, must diversify risks primarily by expanding the pool of insured individuals. For these reasons, it is necessary to determine independent, stable, and sustainable financing sources for the dedicated fund for rare diseases. It is recommended to consider a combination of multiple funding channels, including central government fiscal appropriations, revenues from welfare lottery funds, and donations from philanthropic organizations, to build a national pool for the dedicated fund for rare diseases.

The initial scale of funding may be estimated based on the number of diseases included in the National Rare Disease Catalog, the registered number of patients in the national rare disease direct reporting system, and the costs of drugs that have been approved but not yet included in the NRDL or the CHIIDL, combined with a fixed annual cap on individual out-of-pocket expenses. Building on this, the initial scale of funding and coverage scope for the national rare disease fund can be determined by considering the central government's expenditures, revenue, and budget allocation. Assessment on the scale of funding needed for the dedicated fund on rare disease may be conducted on the principle of determining coverage based on revenue, operating according to the available resources, and gradual implementation.

Implementing a Standardized Application and Review Mechanism and Clarifying Drug Selection Criteria: The dedicated national fund for rare diseases should primarily provide coverage for rare disease drugs used to treat conditions listed in the National Rare Disease Catalog that have not yet been included in the NRDL. An expert committee should be established to evaluate medicines submitted by companies based on a value assessment framework specific to rare diseases, giving priority to drugs that meet the following criteria:

- Drugs treating pediatric rare diseases
- Drugs treating diseases that are life-threatening or severely impair quality of life
- Innovative drugs that address unmet clinical needs
- Drugs with potential long-term benefits and/or high social value

Since rare diseases affect small patient populations, it is difficult to conduct large-scale clinical trials for rare disease drugs or accurately assess the effectiveness of rare disease drugs based solely on pre-market clinical trial data. Authorities may consider implementing a more flexible value assessment framework for the dedicated fund. This framework would accept data from small-sample or single-arm trials when evaluating drugs' eligibility for coverage by the dedicated fund.

Once products have been included in the dedicated fund's scope of coverage, companies could be required to conduct post-marketing real-world studies to generate evidence for post-access value reassessment and coverage renewal negotiations.

Establishing Risk-Sharing Mechanisms Through the Adoption of Innovative Payment Agreements, While Setting a Cap on Individual Out-of-Pocket Expenses: Annual treatment costs for rare disease drugs covered by the dedicated national fund for rare diseases are relatively high. Therefore, the dedicated fund should negotiate innovative payment agreements with pharmaceutical companies and establish risk-sharing mechanisms. China could draw on international and domestic best practices, real-world clinical needs, expert committee recommendations, and the revenue and expenditures of the dedicated fund, when conducting such negotiations. At the same time, planning for the dedicated fund could draw from the Zhejiang practice by setting a cap on individual out-of-pocket expenses. Innovative payment agreements, combined with a cap on individual out-of-pocket expenses, will effectively alleviate the financial burden of high treatment costs for rare disease drugs, enhance the sustainability of the fund, and keep patient medical expense burden within an affordable range.

The dedicated fund could set an annual limit on total reimbursable expenditures for rare disease drugs that fit the following criteria: having a clearly defined patient population, approved in China, addressing significant clinical needs and demonstrating well-established efficacy. The annual limit may be determined based on price negotiations with companies. This annual limit model would facilitate budgeting and expenditure management for the dedicated fund, stabilize companies' price expectations, and ensure reliable supplies of drugs.

The fund may enter into financial outcome-based innovative payment agreements with companies for new rare disease drugs that fit the following criteria: undefined patient populations, unclear clinical efficacy, and uncertain financial impact on the fund. The financial outcome-based agreement would address the considerable operational challenges posed by clinical-outcome-based agreements in designing

study, analyzing data, and refunding patients. The financial outcome-based innovative payment agreements would specify the intended reimbursement price and risk-sharing arrangement. It would also define details such as the cycles, proportions, and conditions for reimbursement, as well as other related terms. In addition, drawing on Italy's best practices, the fund should sign data-collection agreements with pharmaceutical companies, specifying the responsibility of all parties in the data collection process. Under these agreements, companies would bear the costs associated with real-world data collection. The data obtained would provide robust evidence of actual clinical benefits and risks in real-world use, thereby supporting future participation of these drugs in NRDL negotiations.[53]

Establishing Coordination Mechanism with BMI and Complementarity Across the MTMSS: As an integral part of the MTMSS, the national dedicated fund for rare diseases could design effective mechanisms to coordinate with the BMI. This would enable dynamic management of the dedicated fund's list of covered drugs and ensure effective coordination between the delisting of drugs from coverage by the dedicated fund and their inclusion in the NRDL. By effectively integrating the dedicated fund with the NRDL, continuous coverage can be provided for rare disease patients throughout their life-cycle, thereby consistently reducing their overall disease burden.

For drugs included in the coverage of the dedicated fund for the first time, the fund could sign innovative payment agreements with companies for a period of no more than five years. Real-world data collection could be initiated in parallel. During the agreement period, the collected data will be subject to ongoing monitoring and interim analyses. After the agreement expires, a comprehensive assessment may be conducted based on accumulated evidence of the drug's clinical value and its impact on the dedicated fund's budget. Following these assessments, renewal negotiations may be initiated with the manufacturer. Alternatively, drugs marketed as generics

[53] Flick, E. Dawn, Howard R. Terebelo, Susan Fish, Amani Kitali, Vrinda Mahajan, Melissa Nifenecker, Kristen Sullivan, Paul Thaler, Sarah Ussery, David L. Grinblatt. "The Value of Pharmaceutical Industry-Sponsored Patient Registries in Oncology Clinical Research." *The Oncologist*, vol. 28, no. 8, 2023, pp. 657-663. https://doi.org/10.1093/oncolo/oyad110.

or other competing products may seek inclusion in BMI through the NRDL negotiation process.

(II) Under National Guidance, Regions with Relatively Well-Developed Rare Disease Diagnosis, Prevention and Treatment Systems Could Carry-Out Pilots to Accumulate Replicable and Scalable Experiences for the Nationwide Rollout of the Dedicated Fund:

As top-level design and preparatory work for the national dedicated fund progresses, pilot programs could be launched in regions with relatively well-developed rare disease diagnosis, prevention and treatment systems.

Construction of a national dedicated fund could draw from previous efforts in medical insurance policy reform. For example, during the reform of medical insurance payment methods, as national diagnosis-related group (DRG) payment systems were being developed, regional governments actively explored these reforms in parallel. These regional efforts accumulated valuable experience, providing a solid foundation for the subsequent development of national technical standards, while enabling potential risks and challenges to be identified early.

Similarly, in the process of building a national dedicated fund for rare diseases, local pilot programs can be launched in parallel to explore and trial different aspects of national planning, such as fund size estimation, real-world data collecting on drug use cases, and innovative payment agreement arrangements. Local pilot programs would help to identify operational issues and generate practical, scalable experience, effectively controlling risks that may arise during nationwide rollout. This approach would enhance the scientific rigor and feasibility of the national dedicated fund design, while ensuring that the fund is both aligned with China's national conditions and objectively reflects China's rare disease diagnosis and treatment realities.

VI. Conclusion

Improving the medical insurance system for rare diseases is a key task for advancing the "Healthy China" initiative and achieving the goal of health for all. China has already developed a systematic approach to strengthening rare disease

management and upgrading rare disease diagnosis and treatment capacity. China has accelerated the market entry of innovative rare disease drugs through a range of measures and laid a solid foundation for the establishment of the MTMSS. However, clear shortcomings remain in the current system's capacity to finance and provide coverage for high-cost rare disease drugs. Insufficient coverage for high-cost rare disease drugs has become a critical bottleneck, constraining both health equity for rare disease patients and the virtuous cycle of pharmaceutical innovation.

In the next development stage of the 15th Five-Year Plan period, China should establish, under national-level coordination and with local pilot exploration, a dedicated fund for rare diseases to enhance the coverage of high-cost rare disease drugs, thereby strengthening the MTMSS for rare diseases. This would not only substantially reduce patients' disease burden and prevent households from falling into or returning to poverty but would also provide more reasonable returns on investment for innovative pharmaceutical companies. This would advance the development of the innovative drug industry and foster new competitive advantages for industrial development during the 15th Five-Year Plan period.

The institutional designs and policy recommendations proposed in this report are intended to inform relevant authorities as they plan and advance improvements to the rare disease medical insurance system during the 15th Five-Year Plan period. As top-level design is progressively implemented and policy instruments are further refined, China is well positioned to develop a "China model" for a multi-tiered medical security system for rare diseases. Such a system would demonstrate to the global society the unique strength of Chinese institutions, while contributing "Chinese wisdom" to the realization of visions outlined in the *WHA Resolution.*