Your medical records will be kept at the hospital, and researchers, study authorities, and ethics committees will be granted access to your medical records. Any public report of the results of this study will not disclose your personal identity. We will make every effort to protect the privacy of your personal medical data to the extent permitted by law.

Do I have to participate in the study?

Participation in the study is completely voluntary and you may decline to participate or withdraw from the study at any time during the study without affecting your physician's care. If you decide to withdraw from this study, please contact your doctor. You may be required to undergo relevant tests, which can be beneficial to protect your health.

If you have any questions concerning your personal rights and interests, please contact the Ethics Committee of the Court at 025-68306360.

Subject Statement: I have read the above introduction about this study and fully understand the risks and benefits that may arise from participating in this study. I volunteered for this study. I will obtain a signed and dated copy of this informed Consent.

I consent ☐ **or refuse** ☐ use of my medical records and clinical specimens in connection with this study for other studies.

Subject's signature: [redacted] Data: 2022.7.1

Telephone:: _____ Mobile phone: [redacted]

Guardian's Signature: _____ Data: _____

Telephone:: _____ Mobile phone: _____

Investigator statement: I confirm that I have explained to the subject the details of the study, especially the risks and benefits that may arise from participating in the study, and answered all relevant questions of the subject. The subject consented to participate in the study voluntarily. This informed consent is made in duplicate, with one signed one for both the investigator and the subject.

researcher's Signat[redacted]

Telephone:: [redacted]

Informed consent

Dear Sir/Madam,

We invite you to participate in the research on "3M Syndrome" approved by Jiangsu Provincial People's Hospital. Yin Guoyong was in charge of this study. Subjects volunteered to participate in this study, and our center planned to enroll 1 subject. This study has been reviewed and approved by the Ethics Committee of the First Affiliated Hospital of Nanjing Medical University (Jiangsu Provincial People's Hospital).

Why was this study conducted? (Brief introduction of research background and purpose)

3M syndrome is a rare autosomal recessive genetic disorder characterized by severe intrauterine and postpartum developmental delays. At present, there is a lack of effective diagnosis and differential diagnosis techniques. The aim of this study was to extend the molecular spectrum of 3M syndrome.

Who should (or should not) participate in the study? (Inform the main inclusion and exclusion criteria)

Inclusion criteria (excerpt): Suspected 3M syndrome.

What do I need to do if I participate in the study? (mainly to provide detailed information on the research methods, procedures, steps and precautions)

If you are willing to participate in this study, you may be able to further identify the cause. We will conduct a blood test for you when you accept to participate in the study. We will conduct a whole exon sequencing test in New Hope Company and report the results. The test fee will be paid by the researcher, and the samples will be destroyed upon completion of the test and we promise that the samples will not be used for other purposes. You will be followed up within 4 weeks after the examination, and you will need to spend 2 hours to participate in the study. In the above tests, whole exon sequencing is an investigational test (i.e., you do not need to undergo the test if you are not participating in the study).

What are the benefits of participating in a study? (Objectively inform subjects of their direct or indirect benefits and social benefits)

Your condition may be improved by participating in this study, which may help in the diagnosis of 3M syndrome.

What are the risks of participating in a study? (Inform all adverse reactions of experimental drugs and control drugs or the risks and countermeasures of experimental devices and therapies)

Venous drawing risks may include pain and/or ecchymosis and, in rare cases, needle fainting and infection at the acupuncture site, which often resolves on its own. We will observe the possible side effects/adverse reactions through regular inspection, and take necessary measures to prevent and cure them (if necessary, give risk prevention and control guidance, such as hypoglycemia treatment). If you experience any discomfort or adverse reactions, please contact the study physician. In addition, any diagnosis is likely to be missed, and the disease may continue to develop because of missed diagnosis or because of complications with other diseases.

Do I have to pay for the study? (Specify the compensation plan and amount)

In order to compensate you for the inconvenience caused by your participation in the study, the study will pay the cost of the genetic test performed during your participation in the study and the registration fee for follow-up visits.

Is personal information confidential?

受试者声明：我已经阅读了上述有关本研究的介绍，对参加本研究可能产生的风险和受益充分了解。我自愿参加本研究。我将获得一份签署姓名和日期的本知情同意书副本。

我同意☐ 或拒绝☐ 其他研究利用我的与本研究相关的医疗记录和临床标本。

受试者签名：[redacted] 日期：2022 年 7 月 1 日

受试者的联系电话：[redacted] 手机号：[redacted]

（适用时）监护人签名：_____ 日期：__ __ __ __ 年 __ __ 月 __ __ 日

监护人联系电话：_____ 手机号：_____

研究者声明：我确认已向受试者解释了本研究的详细情况，特别是参加本研究可能产生的风险和受益，并回答了受试者的所有有关问题，受试者是出于自愿同意参加本研究。此知情同意书一式两份，研究者与受试者各留一份[redacted]

研究医生签名：[redacted] 日期：2022 年 7 月 1 日

研究医生的工作[redacted] 手机号[redacted]

知情同意书

尊敬的先生/女士：

我们邀请您参加江苏省人民医院批准开展的“3M 综合征”课题研究。本研究由殷国勇负责，受试者自愿参加，本中心计划入组 1 名受试者。本研究已经得到南京医科大学第一附属医院（江苏省人民医院）伦理委员会的审查和批准。

为什么要开展本项研究？（研究背景和目的简介）

3M 综合征是一种罕见的常染色体隐性遗传病，以严重的宫内和产后发育迟缓为特征。目前缺乏有效的诊断和鉴别诊断技术。本研究的目的是扩展 3M 综合征的分子谱。

哪些人适（不）宜参加研究？(告知主要的入选标准及排除标准)

入选标准（节选）：疑似 3M 综合征。

如果参加研究，需要做什么？（主要是详实告知研究方法、过程、步骤以及注意事项）

如果您愿意参加本项研究，您将有可能性进一步明确病因。我们会在您接受参与研究时对您进行一次抽血检查，于新希望公司进行全外显子测序检测，并报告结果，检测费用由研究者支付，样本检测完即销毁并承诺样本不做他用。并会在检查结束后 4 周内对您进行随访，您参加此项研究需花费 2 小时。在上述检查中，全外显子测序是研究性的检查（即，如果您不参加本研究，就不需要接受该检查）。

参加研究有哪些好处？（客观告知受试者本人直接或间接获益以及社会效益）

参加本项研究，您的病情有可能获得改善，本项研究有助于 3M 综合征的确诊。

参加研究有哪些风险？（告知所有试验用药、对照用药的不良反应或试验器械、试验疗法的风险及应对措施）

静脉抽血风险可能有疼痛和/或瘀斑，罕见的情况下，可能会出现晕针，在针刺部位出现感染，但常会自行痊愈。我们将通过定期检查、观察可能引起的副作用/不良反应，并采取必要措施加以防治（必要时给予风险防控指导，如低血糖处理等）。如果出现任何不适和不良反应，请及时与研究医生联系。此外，任何诊断都可能出现漏诊的情况，以及因漏诊或者因合并其他疾病等原因而导致病情继续发展。

参加研究需要支付有关费用吗？（具体说明补偿计划和金额）

为了补偿您参加本研究可能给您带来的不便，本研究将支付您参加本项研究期间所做的基因检查费用以及随访时的挂号费。

个人信息是保密的吗？

您的医疗记录将保存在医院，研究者、研究主管部门、伦理委员会将被允许查阅您的医疗记录。任何有关本项研究结果的公开报告将不会披露您的个人身份。我们将在法律允许的范围内，尽一切努力保护您个人医疗资料的隐私。

我必须参加研究吗？

参加本项研究是完全自愿的，您可以拒绝参加研究，或在研究过程中的任何时间退出本研究，这都不会影响医生对您的治疗。如果您决定退出本研究，请与您的医生联系，您可能被要求进行相关检查，这对保护您的健康是有利的。

如您有涉及个人权益方面的问题可与本院伦理委员会联系，联系电话：025-68306360。