Enabling safe and timely patient access to transformative therapies

Recommendations for Action in the EU Pharmaceutical Strategy

European Alliance for Transformative Therapies

Opportunities and challenges in the patient pathway to cell and gene therapy

Recent years have seen a number of innovative therapies becoming available to patients in Europe. These 'transformative therapies' bring new hope for the treatment of diseases, with the promise that they not only manage the symptoms of severe, disabling or life-limiting conditions but transform and save lives.

These therapies, also known as Advanced Therapy Medicinal Products (ATMPs), are medicinal products based on cells, genes and/or tissue, which are used to regenerate, augment or replace human cells and tissues, to replace or over-ride a defective gene, or to activate healing processes within the body.[1]

The number of therapies in development is increasing very rapidly. It is anticipated that there will be 5 to 10 new ATMPs approved every year as of 2025,[2] bringing the prospect of delivering unprecedented benefits to patients and society. However, for this prospect to materialise, health systems need to adapt their policies to the new realities and address the challenges that emerge due to the misfit between the established policy paradigms and inherent novel features of these innovations.

Unlike other medicinal products, transformative therapies such as ATMPs address the root cause of the disease and can provide long-lasting benefit, and potentially a therapeutic cure, to many people living with severe diseases with just one or few administrations. For instance:

- Gene therapies can be a transformational treatment or a promise of therapeutic cure for several devastating paediatric rare diseases of genetic origin.
- Malfunctioning cells or damaged tissue can be repaired or replaced by the use of stem cells.
- Genetically engineered cells (CAR-Ts) are used to specifically target and kill cancer cells and tumours.

The rapid pace of scientific knowledge and innovation progress is not aligned with the pace of change in health policies. Many diseases are currently addressed with chronic treatments and reimbursement of costs spread over long periods of time. One-off, potentially curative treatment with ATMPs brings a paradigm shift, creating major hope for patients but also challenges for health systems and health budgets in the short and medium term. The adoption of ATMPs also often requires new practices and skills by healthcare providers.

The European Alliance for Transformative Therapies (TRANSFORM) was created to foster effective dialogue around cell and gene therapies and provide evidence-based policy recommendations to enable safe and timely patient access to these new therapies whilst ensuring the sustainability of healthcare systems. This multi-stakeholder Alliance connects Members of the European Parliament (MEPs) and policy-makers with patient groups, medical experts and associations, scientists, researchers, industry actors, networks and other relevant stakeholders.

The European Commission's initiatives in the area of health and pharmaceuticals provide the opportunity to improve the patient access pathway from diagnosis through to reimbursement. Outlined below are recommendations for policy action developed by stakeholders who are experts in the field.

[1] See the definition by the European Medicines Agency: https://www.ema.europa.eu/en/human-regulatory/overview/advanced-therapy-medicinal-products-overview ; For detailed definitions of the different groups of ATMPs, refer to Regulation (EC) No 1394/2007 and Directive 2001/83/EC.
[2] (1) Quinn et al. Value in Health 2019;22(6):621-626. (2) G. Rasi's oral communication, October 2020.

Priorities for Action

1. Create a patient-centric, innovation ecosystem to address unmet medical needs

2. Safeguard patient access by defining appropriate regulatory requirements for ATMPs

3. Enable the use of real-world patient data through the creation of a European Health Data Space

4. Improve infrastructure and enable cross-border patient access to transformative therapies

5. Increase patient access and ensure sustainability of healthcare systems through innovative payment models

6. Share best practices and recommendations on genetic testing and diagnostics

Recommendations for patient access to transformative therapies

Research

Stimulate translational research to ensure that universities, research centres and industry can create partnerships to translate innovation into new patient treatments

Encourage collaborative approaches to identify research priorities and effective incentives to promote innovation in areas of high unmet medical needs

Provide Horizon Europe funding to promote cross-disciplinary and patient-centred education, covering innovative paediatric research

Clinical Development

Reinforce more patient-oriented design, planning and conduct of clinical trials

Review requirements for therapies that contain or consist of GMOs

Promote early dialogue with both regulatory agencies and payers to better design clinical trials and collect relevant data for more streamlined authorisation, pricing and reimbursement and faster access to patients.

Regulatory Approval

Ensure consistent and harmonised implementation of classification criteria of blood, tissues and cells to avoid divergent interpretations and decisions across the Member States

Introduce EU regulatory guidelines, premised on a patient-centric approach, **setting minimum standards on when and how treatment should be offered under the hospital exemption**

Create European registries that ensure transparency by **tracking the use of hospital exemption**

Clarify the interplay of the Clinical Trial Regulation and the Regulations for Medical Devices and for In Vitro Diagnostics

Patient Access

Ensure national authorities **authorise ATMP treatment abroad in an effective and timely manner. Accelerate** reimbursement processes for cross border treatment of patients through the revision of the Cross-border Healthcare Directive and use of the Social Security Regulations

Enhance information and infrastructure for the provision of ATMPs through centres of excellence; Provide clear information on which centres in the EU are administering cell and gene therapies

Disseminate best practices on innovative payment models that can support national governments and spread cost

Enabled by Real-World Data

Ensure the European Health Data Space enables the use of Real-World Evidence (RWE) in the lifecycle management of ATMPs

Enable better coordination between regulators and payers on the use of RWE, and agreed methods for RWE

Ensure better physician and patient involvement in the collection of RWE to improve outcomes and ensure safety

Promote the creation and harmonisation of patient registries to monitor long-term effects and follow-up of ATMPs

Genetic testing and diagnostics

Promote cross-country collaboration through networks and research infrastructures to facilitate sharing of best practices on genetic testing and diagnostics

1. Create a patient-centric, innovation ecosystem to address unmet medical needs

There is considerable scope to facilitate all stakeholders working together to identify research priorities, clarify trial design, and, generally, increase patient engagement throughout research.

Continuing commitment to break down the silos and stimulate translational research is needed to ensure that universities, research centres and industry have the right capabilities and know-how to innovate and create the necessary partnerships to translate innovation into new patient treatments.

Better identification of unmet medical needs will help national/public health systems to identify health priorities in a more coordinated way and will facilitate investment for research in these disease areas, as well as development, approval and adoption of therapies that address them.

Incentives for product development should be effective in promoting innovation in Europe, especially in areas of high unmet medical needs, which can be addressed by transformative therapies. Europe's attractiveness for the development of transformative therapies is critical to ensure that European patients can get timely access to these treatments.

Recommendations

Stimulate translational research to ensure that universities, research centres and industry can create the necessary partnerships to translate innovation into new patient treatments.

Encourage collaborative approaches to identify research priorities and effective incentives to promote innovation in areas of high unmet medical needs.

Enable patient organisations and patient representatives to co-drive the process formally.

Relevant EU Initiatives

- European Industrial Strategy
- Pharmaceutical Strategy for Europe
- Revision of the EU general pharmaceuticals legislation
- Evaluation of the Orphan and Paediatric legislation
- Horizon Europe

Relevant Stakeholder Initiatives

- European Expert Group for Orphan Drug Incentives – Recommendations for Actions
- European Rare Disease Research Coordination and Support Action (ERICA)[3]
- International Rare Diseases Research Consortium (IRDiRC)[4]
- European Paediatric Translational Research Infrastructure – Paediatric Research Manifesto[5]
- ACCELERATE International Multistakeholder Platform – Paediatric Strategy Forums[6]

[3] https://eatris.eu/projects/erica-european-rare-disease-research-coordination-and-support-action/
[4] https://irdirc.org/activities/irdirc-publications/
[5] https://eptri.eu/manifesto-for-paediatric-research/

2. Safeguard patient access by defining appropriate regulatory requirements for ATMPs

Reinforce more patient-oriented design, planning and conduct of clinical trials

An effective way for patients to access transformative therapies is to be in clinical studies designed to respond to their needs. The role of patients can be crucial in developing ATMPs better focused on the patients' needs. This includes addressing practical issues, particularly for younger patients who often have to drastically change their daily lives.

Randomised and placebo-controlled clinical trials, which constitute the norm for the approval of conventional medicinal products, are often not applicable in the case of ATMPs. This is because of the indication being studied, the small patient population (e.g. rare diseases, paediatric population) and the intrinsic characteristics of the product (e.g. autologous ATMP). Innovative trial designs, which are better adapted to the characteristics of transformative therapies and better capture outcomes that are important to patients, are required. To ensure greater access to these therapies, it is critical that trial sponsors engage early on not only with regulators but also with national payers, so that trial designs and results can feed into the economic understanding and evaluation of these novel therapies.

The EU Clinical Trial Regulation,[7] due to be implemented in early 2022, will provide common and harmonised rules for conducting clinical trials in the different EU countries. The effective implementation of this Regulation will be an important step to ensure a more consistent approach to clinical studies, including for trials with innovative design and technologies. Additionally, it will be important to ensure clarity on the interplay between the Clinical Trial Regulation and the new Regulations for Medical Devices (MDR)[8] and for In Vitro Diagnostics (IVDR).[9] As medical device and in vitro diagnostic approvals lie outside the Clinical Trial Regulation, ensuring timely and streamlined approvals will be necessary.

Recommendations

- Reinforce more patient-oriented design, planning and conduct of clinical trials.
- Clarify the interplay of the Clinical Trial Regulation and the Regulations for Medical Devices and for In Vitro Diagnostics.

Relevant EU Initiatives

- HTA Regulation
- Pharmaceutical Strategy for Europe
- Revision of the EU general pharmaceuticals legislation
- Implementation of the EU Clinical Trial Regulation, and implementation of the Medical Devices Regulation (MDR) and In-Vitro Diagnostics Regulation (IVDR)

Relevant Stakeholder Initiatives

- SIOP Europe Essential Medicines and HTA Evaluation Project
- SIOPE Clinical Research Council (SIOPE CRC)[10]

[6] https://www.accelerate-platform.org
[7] Regulation (EU) No 536/2014
[8] Regulation (EU) 2017/745
[9] Regulation (EU) 2017/746
[10] https://siope.eu/european-research-and-standards/clinical-research-council/

Review requirements for therapies that consist of or contain GMOs

Most cell and gene therapies currently in development meet the definition of Genetically Modified Organisms (GMOs) and need to comply with the GMO legislation in Europe.[11;12] Accordingly, cell and gene therapies containing or consisting of GMOs must conform to complex rules on the environmental risk assessment that are not primarily intended for medicinal products. These rules vary greatly across the Member States and lead to divergent decisions depending on the Member State.

> *For the delivery or manufacture of cell and gene therapies, a transporting vector (a virus) is altered to make it non-pathogenic or incapable of replication; if excreted and/or released, the GMO impact on the environment is negligible.*

Such complexity, and the associated formal assessment, leads to delays in patients' access to cell and gene therapies and acts as a deterrent for clinical trial sponsors to conduct their trial in Europe. The revision of the pharmaceutical legislation provides an opportunity to meaningfully simplify the environmental assessment of cell and gene therapies drawing from the experience gained with the temporary exemption for Covid19 vaccines and treatments to comply with GMO requirements. More specifically, the European Commission could work with Member States to streamline the assessment of GMO dossiers for clinical trials and early access programs, and also to develop harmonized templates across the EU.

Recommendations

Review requirements for therapies that consist of or contain GMOs; Simplify the environmental assessment of cell and gene therapies, drawing from experience gained with the temporary exemption for Covid19 vaccines.

Relevant EU Initiatives

- Revision of the EU general pharmaceuticals legislation
- Pharmaceutical Strategy for Europe

[11] Directives 2001/18/EC and 2009/41/EC
[12] The environmental or biosafety risk with gene therapies using non-pathogenic and replication-incompetent vectors, with *in vivo* gene therapies or with products manufactured using genome-editing techniques is negligible.

Bring clarity and consistency around classification and regulatory requirements

The distinction between blood, cells, tissues, ATMPs and medical devices or between the different types of ATMPs (gene therapy, somatic cell therapy, tissue-engineered product or combined product) is not always straightforward and requires careful examination.[13] Currently, the classification from the EMA's Committee for Advanced Therapies (CAT) does not necessarily prevail over the advice from national bodies (e.g. the national medicines agency, or health authorities in charge of medicinal products, blood or cells and tissue), bringing confusion about the requirements for the manufacturing, control, development and use of such products.[14]

The enforcement of common regulatory standards across Europe with clear classification criteria that apply irrespective of the manufacturer (hospital, academic centre or industry) or EU Member State and that are convergent with the standards and classification criteria used by non-EU regulatory bodies (USA, Japan, etc.) are necessary to ensure high-quality healthcare and patient safety in Europe.

It is therefore important to have a mechanism at EU level that avoids any possibility of divergent interpretations and decisions across Member States. A lack of consistency and predictability on classification deters the development of and access to transformative therapies in Europe.

Recommendations

Ensure consistent and harmonised implementation of classification criteria of blood, tissues and cells to avoid divergent interpretations and decisions across Member States.

Relevant EU Initiatives

- Revision of the EU legislation on blood, tissues and cells[15]
- Pharmaceutical Strategy for Europe
- Revision of the EU general pharmaceuticals legislation

[13] The currently applicable European legislation defines when a tissue, cell or gene-based product needs to be considered as an ATMP, i.e. when such product has been subject to a substantial manipulation (e.g. a cell culture expansion) and/or when they are used in a non-homologous way (i.e. cells or tissues that are not intended to be used for the same essential function(s) in the recipient and the donor, e.g. adipose cells manipulated and used to repair bone defects). Cells, genes and tissues can also be used in combination with scaffold or other medical devices.

[14] The European Commission's evaluation report acknowledges the issue: https://ec.europa.eu/health/blood_tissues_organs/policy/evaluation_en

[15] "Blood, tissues and cells for medical treatments & therapies" European Commission Public Consultation https://ec.europa.eu/info/law/better-regulation/have-your-say/initiatives/12734-Revision-of-the-Union-legislation-on-blood-tissues-and-cells

Ensure regulatory requirements are clarified and stakeholders are aligned

The hospital exemption (HE) provides a useful pathway for patients with severe medical conditions to receive an ATMP under controlled conditions in cases where no authorized medicinal product is available. Investigational treatments offered under hospital exemption are often the last resort for patients. However, the hospital exemption is interpreted and implemented differently across EU Member States.[16]

TRANSFORM believes that a patient-centric approach must be taken to define when and how a treatment should be offered under hospital exemption. Transparency about the use of the hospital exemption in the different Member States is important to ensure that it is used only where no registered products are available. The hospital exemption should be subject to consistent regulatory oversight across EU Member States requiring the evaluation of minimum quality, preclinical, and possibly clinical data before an authorization is granted. Moreover, the hospitals providing ATMPs under hospital exemption should be held accountable for minimum standards in terms of capabilities of equipment, staff, reproducibility, good manufacturing practice (GMP). An EU regulatory guideline setting these minimum standards of when and how a treatment should be offered under hospital exemption would be helpful.

To ensure patient safety, a registry should be established with publicly accessible information related to ATMPs produced under hospital exemption. The hospital exemption pathway should not be used to bypass the process of seeking a marketing authorization, which ensures a product's quality, safety, and efficacy. When a treatment receives marketing authorization, patients should be given access to such treatment. Where the use of an experimental ATMP under hospital exemption becomes routine, the provider should be required to run a clinical trial and seek marketing authorization to make the treatment accessible to more patients.

The use of unregulated products (e.g. stem cells) in some clinics, with no hospital exemption license and no regulatory oversight, promising a wide range of benefits using poorly characterized medicinal products with little evidence of effectiveness and no quality control, is of great concern since it could harm patients. Increased awareness amongst academics and hospitals of the requirements for ATMPs, increased regulatory oversight and increased control of such practices, including when such use is carried out under a national hospital exemption framework, should be enforced.

Recommendations

Introduce EU regulatory guideline, premised on a patient-centric approach, setting minimum standards of when and how a treatment should be offered under hospital exemption.

Create European registries to seek transparency around the hospital exemption with details of the products, hospitals, their uses/indications, number of patients intended to be treated, duration of the hospital exemption authorisations, etc.

Relevant EU Initiatives

- Pharmaceutical Strategy for Europe
- Revision of the EU general pharmaceuticals legislation
- Revision of Paediatric and Orphan Regulations

Relevant Stakeholder Initiatives

- RARE IMPACT[17]

[16] The legal basis for hospital exemption is found in article 28 (2) of the ATMP Regulation N°1394/2007
[17] https://rareimpact.eu/

3. Enable the use of real-world patient data through the creation of a European Health Data Space

Real-world data allows the systematic collection of data about a patient's health status or the delivery of care from a variety of sources that goes beyond conventional clinical trials. Such sources include electronic health records, claims, registries and other sources of patient-generated health data such as mobile apps.

Since most transformative therapies are administered only once and can provide long-lasting effects, the collection of real-world data is a critical component in their life-cycle assessment. It facilitates understanding of uncertainties on long-term effect, safety, health-related quality of life and long-term follow up of ATMP treatments. It also provides natural history datasets in key disease areas that can be used for the indirect detection of a product's therapeutic effect when direct comparisons are not feasible.

The creation of a European Health Data Space (EHDS)[18], which would foster the exchange and sharing of different kinds of health data (electronic health records, genomics, registries, etc.) in Europe, is an important step forward to address these challenges. Since real-world data are critically important in the life-cycle assessment of ATMPs, initiatives and pilots under the EHDS project should prioritise these products. Paediatric peculiarities shall be addressed in setting up the legal/regulatory and operative framework.

Recommendations

Ensure the European Health Data Space enables the use of real-world data in the lifecycle management of ATMPs.

Enable better coordination between regulators and payers on the use of real-world data, and agreed methods for real-world evidence (the evidence generated from real-world data).

Ensure better physician and patient involvement in the collection of real-world data to improve outcomes and ensure safety in the long-term.

Relevant EU Initiatives

- European Health Data Space
- EMA's DARWIN EU (Data Analytics and Real World Interrogation Network)
- INSPIRE Knowledge Base (Linked Data Advanced)[19]

Relevant Stakeholder Initiatives

- RWE4Decisions[20]
- Get Real Institute[21]
- RARE IMPACT[22]

[18] https://ec.europa.eu/health/ehealth/dataspace_en
[19] https://inspire.ec.europa.eu/training/linked-data-advanced
[20] https://rwe4decisions.com/
[21] https://www.getreal-institute.org/
[22] https://rareimpact.eu/

4. Improve infrastructure and enable cross-border patient access to transformative therapies

Develop the infrastructure for ATMPs & enable cross-border collaboration and care

Due to the complexity of cell and gene therapies in terms of translational research, manufacturing and delivery, a certain degree of infrastructural capacity is required for their proper provision. For this reason, TRANSFORM recommends that these complex therapies are delivered in qualified centres with the necessary facilities and specialist knowledge on both these innovative therapies and the conditions they intend to treat.

The expertise and facilities required to provide cell and gene therapies place additional requirements on healthcare providers. These include site certification processes, which are complex, time-consuming and resource-intensive, and they may divert scarce resources away from providing care. EU Member States should leverage and further build on the work carried out in rare diseases and promote the distribution of these novel therapies through rare disease centres of reference or centres of excellence,[23] including through improved national funding. Clarity is needed on which centres in the EU are administering cell and gene therapies. Certification or other ATMP-administration designation processes should involve all relevant stakeholders, including specialist clinicians, patient representatives, hospital managers, government officials and others at European and/or national levels.

Cell and gene therapies require formal training before healthcare professionals have the adequate information to convey the benefits and risks to patients and can administer the treatments. Additional specific training should be required for healthcare professionals in charge of paediatric patients. Post-treatment follow-up care should also be carefully planned and budgeted, including adequate psycho-social support by specialists. The launch or tailoring of European funding programmes (e.g. Horizon Europe) is needed to promote cross-disciplinary and patient-centred education for healthcare professionals on cell and gene therapies.

Recommendations

- Leverage existing centres of reference/excellence for the delivery of ATMPs.
- Encourage multi-stakeholder discussions to designate suitable centres and develop adequate delivery and follow-up care protocols that support and follow the patient long after the delivery of the therapy.
- Consider enhancing the information and infrastructure for the provision of ATMPs through centres of excellence. Provide clear information on and/or designation of which centres in the EU are administering cell and gene therapies.
- Provide Horizon Europe funding to promote cross-disciplinary and patient-centred education, covering sectors currently underrepresented such as paediatric innovation.

[23] See for example Advanced Therapy Treatment Centres network (UK) - https://www.theattcnetwork.co.uk

Enable cross-border delivery and reimbursement of treatments for patients

ATMPs are highly personalised treatments with complex manufacturing and distribution processes. Due to their complex nature, the need for trained healthcare providers and/or the rarity of the condition they aim to address, many of these therapies will be available only in a small number of treatment centres in Europe. As a result, many patients in Europe may need to travel cross-border to benefit from treatment with a transformative therapy.

The European legislation provides two different routes for patients to access healthcare abroad and have the costs covered by their national health service/health insurance provider under European law: the Social Security Regulations[24] and the Directive on patients' rights in cross-border healthcare[25]. However, in practice, neither of these routes actually work in the case of treatment with transformative therapies:

- The **Cross-Border Healthcare Directive** requires upfront payment from patients for their treatment, with reimbursement limited to what is authorised in their home country, something that is neither feasible nor practical in the case of ATMPs with high upfront costs.
- The **Social Security Regulations** require a pre-authorisation by the national health service/health insurance in the patient's home country, something that creates burdens: travel and accommodation costs incurred by patients and their families are not reimbursed; the need for the payer's pre-authorisation in the country of origin can potentially slow down or even prevent access to life-saving and other urgent healthcare treatments; there may be difficulties when the ATMP is not included in the basket of care in the home country.

Urgent actions are needed to address these shortcomings and ensure that patients can travel within the European Union for access to transformative therapies taking into account age-related needs which are different for children, a population that has been neglected and insufficiently considered.

Recommendations

Ensure national authorities authorise ATMP treatment abroad in an effective and timely manner.

Closer cooperation between the ERNs to set up simple and standardised care pathways and adequate support to navigate national and cross-border healthcare pathways.

Accelerate reimbursement processes for cross-border treatment of patients through the revision of the Cross-border Healthcare Directive and use of the Social Security Regulations.

Relevant EU Initiatives

- European Reference Networks[26]
- Horizon Europe (9th Framework Programme)
- Evaluation of the Cross-Border Healthcare Directive
- Pharmaceutical Strategy for Europe
- Proposed EU HTA Regulation

Relevant Stakeholder Initiatives

- EURORDIS - RARE IMPACT[27]
- EPTRI - Paediatric Research Manifesto[28]
- EAHAD-EHC Joint Statement on Promoting hub-and-spoke model for the treatment of haemophilia and rare bleeding disorders using gene therapies[29]

[24] Social Security Regulations (EC) 883/2004 and 987/2009
[25] Directive 2011/24/EU on patients' rights in cross-border healthcare
[26] https://ec.europa.eu/health/ern_en
[27] https://rareimpact.eu/site/wp-content/uploads/2020/11/RARE-IMPACT-European-Assessment_v1_2020-11-23.pdf
[28] https://eptri.eu/manifesto-for-paediatric-research/
[29] https://eahad.org/wp-content/uploads/2020/05/Hub-and-Spoke.pdf

5. Increase patient access and ensure sustainability of healthcare systems through innovative payment models

To ensure timely patient access to ATMPs, TRANSFORM calls for a coherent approach with the scientific community, treatment centres, payers and manufacturers to define key parameters, criteria and assessment over time to establish outcome-based models, and to increase the use of Real-World Evidence.

Innovative payment models such as outcome-based agreements, conditional reimbursement or annuity-based payment models have been proposed for transformative therapies.[30] For health budgets to absorb their costs they need to be spread over time and/or conditioned with outcome results to account for long-term unknowns and uncertainties.[31]

Whilst pricing and reimbursement of medicinal products fall within national competences, there is a need for a more coordinated approach across Europe, with sharing best practices on innovative payment models and ways to address barriers for their adoption. There is also a need for European coordinated data collection on these treatments to better assess them in real-world settings. These data will provide further insight into the benefits and risks of these novel therapies and support payers' decision-making process. This demonstrates again the importance of patient registries to provide evidence on real-world use.

Furthermore, ATMPs require a future-proofed HTA system. Current discussions around the HTA Regulation show there is a willingness to facilitate some alignment across Member States on the value assessment of new technologies and to find ways to address uncertainties in the longer term, e.g. by defining important outcomes in outcome-based agreements.

Recommendations

Disseminate best practices on innovative payment models that can support national governments to increase patient access to transformative therapies and spread costs.

Coordinate post-marketing data collection and use of registries.

Relevant EU Initiatives

- Proposed HTA Regulation
- Cross-country collaborations such as BeneluxA or FINOSE
- Pharmaceutical Strategy for Europe
- Revision of the EU general pharmaceuticals legislation

Relevant Stakeholder Initiatives

- EURORDIS - RARE IMPACT[32]
- World Federation of Haemophilia (WFH) Global Gene Therapy Registry[33]

30 Cf EUCOPE Position Paper: New Payment and Funding Approaches for A

TMPs https://www.eucope.org/paper-new-payment-funding-approaches-for-atmps/

31 Cf EUCOPE Position Paper: New Payment and Funding Approaches for ATMPs https://www.eucope.org/paper-new-payment-funding-approaches-for-atmps/

32 https://rareimpact.eu/

33 https://news.wfh.org/article-on-the-wfh-gene-therapy-registry-published/

6. Share best practices and recommendations on genetic testing and diagnostics

Many transformative therapies address rare genetic diseases and deliver the greatest benefits when administered early in the patient's life. However, many patients with rare genetic conditions are not diagnosed or diagnosed too late to benefit from therapies that could be life-saving or prevent debilitating effects.

Newborn screening and genetic testing are important to identify and treat patients early, but the adoption of these key tools varies significantly across Member States. Cross-country collaboration through networks and research infrastructures can facilitate sharing of best practices; and recommendations on newborn screening and diagnostics could increase the chances for patients to benefit from life-saving therapies. Such shared efforts as well as better use of European Reference Networks for diagnostic purposes would ensure more equal treatment opportunities across Member States.

Recommendations

Promote cross-country collaboration through networks and research infrastructures to facilitate sharing of best practices.

Relevant EU Initiatives

- European Reference Networks[34]

Relevant Stakeholder Initiatives

- Screen 4 Rare[35]
- European Alliance for Newborn Screening for Spinal Muscular Atrophy (SMA NBS Alliance)[36]
- EURORDIS - RARE IMPACT[37]
- EURORDIS Key Principles for Newborn Screening (January 2021)[38]

[34] https://ec.europa.eu/health/ern/networks_en
[35] https://ipopi.org/newborn-screening-for-rare-diseases-campaign-continues-with-screen-4-rare/
[36] https://www.sma-europe.eu/news/announcing-the-launch-of-the-european-alliance-for-newborn-screening-for-spinal-muscular-atrophy-sma-nbs-alliance/
[37] https://rareimpact.eu/
[38] https://download2.eurordis.org/documents/pdf/eurordis_nbs_position_paper.pdf

About the European Alliance for Transformative Therapies (TRANSFORM)

The European Alliance for Transformative Therapies (TRANSFORM) is a multi-stakeholder Alliance that connects Members of the European Parliament (MEPs) and policy-makers with patient groups, medical experts and associations, scientists, researchers, industry actors, networks and other relevant stakeholders. TRANSFORM aims to foster effective dialogue and provide evidence-based policy recommendations to enable safe and timely patient access to cell and gene therapies, whilst ensuring sustainability of healthcare systems.

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Contributors

EURORDIS
Rare Diseases Europe

EPTRI
European Paediatric Translational Research Infrastructure

SIOP Europe
The European Society for Paediatric Oncology

CCI Europe
Childhood Cancer International Europe

EAHAD
The European Association for Haemophilia and Allied Disorders

EHC
European Haemophilia Consortium

EU-EYE
European Alliance for Vision Research and Ophthalmology

EUCOPE
The European Confederation of Pharmaceutical Entrepreneurs

Please contact secretariat@transformalliance.eu if you wish to endorse the Recommendations and contribute to TRANSFORM's work programme