The Reform of the Newborn Screening Policy: Spinal Muscular Atrophy

Blanka Bartos, Dr. Jur., LLM*

ABSTRACT: One in every 10,000 children is born with SMA and half of them will not even live two years. It is a hereditary genetic disorder, where the muscles die. If it is discovered just after birth, newborns can get the newest medicines to maintain their health. Unlike some other common genetic diseases (e.g. Down-Syndrome), SMA can be screened prior to pregnancy to determine whether the parents are carriers. In Hungary, people have urged reform, due to the baby Zente case, whose story has reached millions. Australia and Germany have also discovered the need for screenings. However, the US has already introduced newborn screening for SMA, far ahead of European countries. National policies should adhere to the same path to contribute to appropriate family planning and to make the treatment available as soon as needed to provide a longer and better life for sick infants.

Keywords: **newborn screening, SMA, family planning, national health policy, human rights**

* Dr. Jur. at the University of Szeged (Hungary, 2017); LL.M. at the University of Toledo (USA) and University of Szeged (Hungary, 2014) in American law; LL.M. at the University Paris 2 Panthéon-Assas (France, 2018) in French, European, and International Business Law; and Ph.D. candidate at the University of Bourgogne Franche-Comté (France) in contracts and international trade law.

Introduction

Newborn screening has been available since the 1960s and has saved lives.[1] Countries have continuously expanded the range of screened diseases prior to a baby's birth including Spinal Muscular Atrophy or SMA. We could be optimistic and patient as health care policy and regulations constantly change in parallel with innovation, however, some countries need to take the first steps, which will motivate others. The USA and Australia have already changed their legislation to dedicate financial resources, technology, and administrative systems for this goal, and now, it is Europe's turn to follow this strategy.

About SMA

Spinal muscular atrophy is caused by a mutation or an absence of the survival motor neuron gene (SMN1). Normally, it produces a protein for the function of the nerves, which control the muscles. Without SMN1, the muscles become weak and eventually die. The disease becomes life-threatening when the basic life support organs (lungs, heart, the digestive system) shut down, nevertheless, the illness does not affect the ability to think, and the patient's mental health remains intact.[2]

We can distinguish SMA 1, 2, 3, 4, based on the age of onset and severity. SMA 1 is the most severe and common type, affecting ca. 60% of SMA patients. It has nine further sub-groups, patients with SMA 1/1 need intensive care to stay alive. The symptoms of SMA 1 appear before the age of six months and without appropriate treatment, the majority of these babies cannot celebrate their second birthday. In contrast to this, the signs of SMA 2 appear between the age of six months - two years and children can reach adulthood, but they will never walk without treatment. SMA 3 is diagnosed from the age of 18 months until the tail end of the teenage years. These children can walk initially, but they lose this ability as they get older. The rarest SMA 4 begins in adulthood. It can appear anywhere from the age of 18 up until 35. There are further mutations of the SMN1 gene which are common in all forms.

SMA can affect any race or gender. It is the number one genetic cause of death among infants in the USA. This illness affects 1 in 11,000 births, and about 1 in every 50 Americans is a genetic carrier. With a DNA test, people can find out whether they are a carrier. If they are thinking about starting a family and both of the partners are carriers, they have a 25% chance that their baby will have SMA. They have several options including no prenatal testing, prenatal testing, adoption, and pre-implantation genetic diagnosis (PGD[3]).

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[1] Caggana, M., Jones, E. A., Shahied, S. I., Tanksley, S., Hermerath, C. A., & Lubin, I. M. (2013). Newborn screening: from Guthrie to whole genome sequencing. Public health reports (Washington, D.C.: 1974), 128 (Suppl 2), pp. 14–19. https://doi.org/10.1177/00333549131280S204

[2] About SMA, Cure SMA https://www.curesma.org/about-sma/

[3] PGD screens embryos for genetic disorders and selects the unaffected embryos for implantation.

Zente Baby

It would be hard to find a person in Hungary who has not heard about Zente, a little boy, who suffered from SMA 1 (I/9) syndrome. In September 2019, the family needed to raise 2.1 million US dollars before the end of the year to buy the newest medicine, Zolgensma.[4] Fortunately, they succeeded in a matter of days. Hungarians donated a sufficient amount of money to order this American drug.[5] Zente was one of the first infants given the drug in Europe and the first one in Hungary.[6] Thanks to this campaign, people received further information regarding SMA disease and became more attentive to infants with this deadly condition.

Zente was born on February 28, 2018, weighing 3470g and measuring 47cm, just like a normal, healthy baby.[7] The delivery happened quickly and safely, the baby got a 9-10 Apgar score[8] (out of 10). Nevertheless, when the family arrived home, problems began to appear. Bowel movements were very difficult and the infant was not able to raise his head. Physiotherapy had positive results, but at the age of 4-5 months, his development plattoed and he was unable to turn onto his stomach. Although the physiotherapist recommended patience, the parents wanted to consult a specialist.[9] The neurologist performed a blood test on Zente, which, unfortunately, came back positive for SMA. The baby boy was fortunate because his genetic illness was revealed at an early stage, but it would not have happened if his mother, Krisztina Tóth, had been more "patient"—as was recommended. Neither the weekly visits by the midwife nor the medical examinations by the pediatrician were enough to recognize the possibility of a genetic disorder. Thanks to his mother's vigilance and consulting a physiotherapy specialist, Zente was diagnosed in time and received the proper treatments.

Nevertheless, the mother does not believe that this is the fault of the doctors, rather the fault of the system. Other mothers with SMA-positive children continue to urge reform by offering a blood test just after birth in order to receive an early diagnosis. The earlier the diagnosis, the greater chances there are to reduce the symptoms of this disease. In addition, there is also more time to collect the money for Zolgensma[10] or to

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[4] Génterápia—a 21. század gyógymódja, Gyógyszertechnológia.hu https://gyogyszertechnologia.hu/genterapia-a-21-szazad-gyogymodja/?fbclid=IwAR2bIBdbzhJzhDp4VJ8iNY4WOkY5VVOZt2WzwyitAtzkouHMIAHcKv8E-M9I

[5] A kétéves kisfiú, aki kis idöre jó hellyé varázsolta Magyarországot, hvg.hu, https://m.hvg.hu/itthon/20191231_A_keteves_aki_egy_kis_idore_jo_hellye_varazsolta_Magyarorszagot?fbclid=IwAR1tca7ANvC52oAva9tlqz15EpU2WV_3_DuS3t_4jRcmcV172QzX3TAoik4

[6] Mínusz 60 fokra lehütve érkezett Zente forradalmi gyógyszere, ö a negyedik európai, aki megkapta, hvg.hu https://hvg.hu/elet/20191029_zente_zolgensma_infzi_bethesda

[7] Zente SMA 1 Tiny Hero . Stronger than SMA, Personal blog, Facebook https://www.facebook.com/Zente-SMA-1-Tiny-Hero-Stronger-than-SMA-350892568989729

[8] Apgar score, MedlinePlus https://medlineplus.gov/ency/article/003402.htm

[9] Dévény, Anna http://www.deveny.hu/

[10] Zolgensma https://www.zolgensma.com/

apply for Spinraza[11] through the government. Although it is not possible to cure SMA, the symptoms can be reduced up to 90% with medication, thus the patient can have a full, long life.[12]

This family has already made a major appeal to the Hungarian state. The Parliament has voted for providing the drug Spinraza to every SMA child under 18 without exception.[13] At that time, Spinraza was the most modern registered medication in the EU[14] (since May 2020, it is Zolgensma[15]). At this time, 54 Hungarian patients may receive this treatment for free, instead of paying approximately 566,000 US dollars out of pocket. Before this decision, only those whose condition was justified received it. The others would have to pay this amount by themselves, however, none of them had sufficient funds to do that.[16] One month after Zente received Zolgensma, the Parliament changed the budget to include Spinraza.[17] If such a short time was enough to increase the budget of this cure, perhaps, the country could devote more sources to the prevention. In the USA, France,[18] and Australia, Spinraza is covered conditionally by health insurance, and unconditionally in Germany.[19] Therefore, we may conclude that Hungary is on a right track for taking better care of SMA patients.

Newborn Screening

In this case, if the parents wish to know prior to birth whether their future child has SMA, they can ask for either a blood test before pregnancy or a prenatal screening after conception.[20] Usually, they have these possibilities when both of the parents know that they are SMA carriers or they have a carrier or case in the family. If they do not have this information, generally, they need to wait until the first symptoms appear to

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[11] Spinraza https://www.spinraza.com/

[12] Danó, Anna: Nem csak a pénzen múlik Zente kezelése, Népszava, https://nepszava.hu/3051959_nem-csak-a-penzen-mulik-zente-kezelese

[13] Minden 18 év alatti gyermek részesülhet Magyarországon a Spinraza kezelésben, Euronews https://hu.euronews.com/2019/11/29/minden-18-ev-alatti-sma-s-beteg-reszesulhet-magyarorszagon-a-spinraza-kezelesben

[14] Ibid.

[15] Zolgensma, EMA , https://www.ema.europa.eu/en/medicines/human/EPAR/zolgensma

[16] A Magyarország 2019. évi központi költségvetéséről szóló 2018. évi L. törvénynek az SMA-betegség gyógyítására kifejlesztett gyógyszer támogatásához szükséges módosításáról szóló T/4668. számú törvényjavaslat (Döntés képviselöi önálló indítvány tárgysorozatba vételéröl), Jegyzökönyv az Országgyülés Költségvetési bizottságának 2019. március 12-én, kedden 10 óra 03 perckor az Országház Széll Kálmán termében (föemelet 64.) megtartott üléséröl, Ikt. sz.: KVB-41/16-8/2019., 15-21.

[17] Minden 18 év alatti SMA-s beteg megkaphatja a kezelést Magyarország Kormánya https://www.kormany.hu/hu/emberi-eroforrasok-miniszteriuma/hirek/minden-18-ev-alatti-sma-s-beteg-megkaphatja-a-kezelest

[18] Accord en France sur le prix du Spinraza, médicament pour une maladie génétique rare, Figaro, https://www.lefigaro.fr/flash-eco/accord-en-france-sur-le-prix-du-spinraza-medicament-pour-une-maladie-genetique-rare-20190418

[19] Reimbursement Information on Spinraza (nusinersen), TheSocialMedWork https://thesocialmedwork.com/blog/reimbursement-information-on-spinraza-nusinersen#:~:text=Reimbursement%20information%20on%20Spinraza%20(nusinersen),-Posted%3A%20January%2018&text=Spinraza%20(nusinersen)%20is%20indicated%20for,5ml%20vial%20for%20one%20treatment.

[20] Testing & Diagnosis, Cure SMA https://www.curesma.org/testing-diagnosis/#newbornscreening

conduct an SMA test.[21] However, if a blood test can be conducted immediately after birth, treatments have better outcomes, thus the baby has more chances to maintain the condition for survival.[22] In evidence that supports this theory, in 2018, 23 American states screened newborns for SMA—in addition to other genetic disorders including endocrine, metabolic, hearing loss, and critical congenital heart defects (CCHDs).[23] A further 13 states have already adopted the program and seek to activate it within two years. Another three states have trials in this field.[24] To succeed, the Advisory Committee must recommend these conditions to the Commissioner of Health, making arguments at the Public Health Council, where they will vote regarding this question.[25] According to the wish of the SMA community, all states will include the SMA test in newborn screening in the future. This test is quite easy to implement. In the baby's first days of life, a few drops of blood are taken onto a special filter paper (Guthrie cards) and then analyzed in a laboratory.[26]

In Australia, as in the USA, SMA testing began in 2018.[27] For the moment, it is only a two-year program, however, it may be continued into the future. According to the results, there have been more than 100,000 newborns screened and 10 of them were positive.[28]

In Germany, two federal states had a one-year pilot project regarding testing SMA on newborns.[29] Bavaria and North-Rhine Westphalia screened 165,525 newborns and 22 SMA cases were detected between January 2018 and February 2019. In conclusion, German officials recommend other countries where SMA treatment is available to do the same. Their motivation was the creation of a new drug for this genetic disease, Spinraza (nusinersen), which was released at the end of 2016.[30] It has historical importance because this drug was the first offered to maintain muscle condition with SMA. This one-year research was not financed by the German government, but by the German Cystinosis Foundation. This is the reason it will not be continued and will not automatically be included in every newborn screening.

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[21] Ibid.

[22] Ibid.

[23] Centers for Disease Control and Prevention, Newborn Screening Portal https://www.cdc.gov/newbornscreening/index.html

[24] Newborn Screening Programs, Cure SMA https://curesma.wpengine.com/newborn-screening-for-sma/#genetictesting State Fact Sheets, Cure SMA

[25] SMA State Fact Sheet, Massachusetts https://curesma.wpengine.com/wp-content/uploads/2020/02/SMA-State-Fact-Sheet_2020_MA_v4.pdf

[26] Guthrie test, Embryology https://embryology.med.unsw.edu.au/embryology/index.php/Guthrie_test

[27] Guthrie test—Australia, Embryology https://embryology.med.unsw.edu.au/embryology/index.php/Guthrie_test#Australia

[28] Kariyawasam, D.S.T., Russell, J.S., Wiley, V. et al. The implementation of newborn screening for spinal muscular atrophy: the Australian experience. Genet Med 22, 557–565 (2020) https://doi.org/10.1038/s41436-019-0673-0

[29] Journal of Neuromuscular Diseases, vol. 6, no. 4, pp. 503-515, 2019

[30] FDA approves first drug for spinal muscular atrophy, U:S: Food & Drug Administration, 23.12.2016. https://www.fda.gov/news-events/press-announcements/fda-approves-first-drug-spinal-muscular-atrophy

In France, which is considered to have one of the best health care systems in the world,[31] 13 illnesses are tested automatically after birth and another 17 will be added by 2023. Unfortunately, SMA is not among them. In 2011, the French National Authority for Health recommended including[32] MCAD deficiency[33] in the screening, and it was finally accepted in 2020.[34] Taking into consideration the progress of this decision in the last nine years, we cannot expect a faster evaluation from the application of the SMA tests either.

In Hungary, newborn screening has been applied since the '70s and has been obligatory[35] since 2007. In 2020, up to 26 metabolic disorders can be tested.[36] This number is better than the majority of the European countries but less than the US where they can screen for more than 40 diseases. The Head of the Hungarian Metabolic Center, László Szönyi declared that it is essential to reveal these illnesses before the symptoms appear. In this regard, they can cure patients immediately without making them suffer irreversible damage to their mental or physical health. Especially since the cure for most of these illnesses is easy to apply, simple, and affordable. Even with dietary changes and taking particular vitamins, they can improve the patient's condition. He (Szonyi) added that a further test against cystic fibrosis would also be beneficial. According to their statistics, newborn screening can save lives, approximately 50 to 60,000 babies a year.[37]

Both Germany and Hungary have aimed to include cystic fibrosis (CF) in newborn screening shortly. It seems that the United States is ahead of them in this field, all 50 states screen for it after birth.[38] Cystic fibrosis is a genetic disease that causes lung infections and inappropriate digestion. There are more than 70,000 registered patients in the world, 30,000 of them being in the US. One thousand more new cases appear each year, 75% of them are diagnosed before the age of two. Although there is still no cure for this illness, treatments can improve the chances of greater longevity.[39]

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[31] Mark Rice-Oxley: Which country has world's best healthcare system? The Guardian, 09.02.2016 https://www.theguardian.com/society/2016/feb/09/which-country-has-worlds-best-healthcare-system-this-is-the-nhs

[32] Recommendations for the expansion of newborn screening to MCAD deficiency, Summary of Public Health Recommendations, HAS, June 2011, https://www.has-sante.fr/upload/docs/application/pdf/2011-07/fs_depistage_neonatal-en-v2.pdf

[33] MCAD deficiency is an inherited metabolic disorder characterised by the inability of the body to use fat. While children with MCAD do not have symptoms at birth, they may develop a metabolic crisis (e.g. during intercurrent illness), which may rapidly lead to coma or death.

[34] Dépistage Néonatal: Quelle maladies dépister? Haute Autorité de Santé (HAS), 03.02.2020. https://www.has-sante.fr/jcms/p_3149627/fr/depistage-neonatal-quelles-maladies-depister

[35] Decree of the Minister of Health in Hungary 44/2007. (IX. 29)

[36] Újszülöttkori szürövizsgálatok, László Szönyi, I.sz. Children's Clinic Budapest http://www.gyermekklinika.semmelweis.hu/upload/seaok1gyermek/document/2009.03.19.sz337nyilszl_jszlttkorisz369r337vizsglatok.pdf

[37] Czétényi, Rita: Évente 50-60 életet ment meg az újszülöttkori szürövizsgálat, Semmelweis Hírek, 28.02.2013 https://semmelweis.hu/hirek/2013/02/28/evente-50-60-eletet-ment-meg-az-ujszulottkori-szurovizsgalat/

[38] Newborn Screening for CF, Cystic Fibrosis Foundation https://www.cff.org/What-is-CF/Testing/Newborn-Screening-for-CF/

[39] About Cystic Fibrosis, Cystic Fibrosis Foundation https://www.cff.org/What-is-CF/About-Cystic-Fibrosis/

We can see from the numbers, why countries are focused on cystic fibrosis rather than SMA. There are more than 40,000 CF cases in Europe, 560 in Hungary.[40] Although the registration of SMA patients does not have official statistics, there are approximately 10 to 25,000 American patients[41] and 120-300 cases in Hungary.[42]

Carrier Screening for SMA

Carrier screening would be more useful than newborn SMA testing, however, it is less utilized. If women who are planning to start a family could have the possibility for a blood test to discover whether they are carriers, they would have more choices. In the case where the woman discovers she is a carrier, her partner could be screened as well.[43] If both of them are carriers and they do not want to risk (25%) that their future child will suffer from SMA, they can choose either in vitro fertilization or intrauterine insemination.[44] If they prefer natural conception, they can also give up the plan of carrying a child and can choose adoption instead. If they opt for natural conception, doctors can test the fetus for SMA by amniocentesis or chorionic villus sampling.[45] The choice of the parents is more difficult in this case; they can prepare in advance for having a child with special needs for a lifetime or in the worst case, with a lower life expectancy. Nevertheless, some parents choose to terminate the pregnancy at an early stage or give up their newborn for adoption.[46]

The implementation of carrier screening would be temporary. If governments made newborn screening obligatory from 2020 forward, individuals would know by birth whether they are carriers. In 40 years, there would be no need for newborn testing, but for the moment, it is quite useful for young adults. Women should be informed about and offered this possibility without additional payment. Currently, in the US, only people with an SMA family history are offered testing.[47] However, it is already a good start as it identifies 2.6% of couples at risk.[48] Since every one in fifty people is a

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[40] European Cystic Fibrosis Society, Patient Registry https://www.ecfs.eu/sites/default/files/general-content-images/working-groups/ecfs-patient-registry/Hungary_Poster_ECFSPR.pdf

[41] SMA Overview, SMA Foundation http://www.smafoundation.org/wp-content/uploads/2012/03/SMA-Overview.pdf

[42] Joób Sándor: Van egy új szupergyógyszer, csak az állam nem adná oda Index, 04.05.2018. https://index.hu/belfold/2018/05/04/gyogyszer_neak_sma_gyogyszertamogatas_betegseg/

[43] Carrier Screening for Spinal Muscular Atrophy, The American College of Obstetricians and Gynecologists https://www.acog.org/patient-resources/faqs/pregnancy/carrier-screening-for-spinal-muscular-atrophy

[44] Norrgard, K. (2008) Medical ethics: genetic testing and spinal muscular atrophy. Nature Education 1(1):88

[45] Ibid.

[46] Prior, Thomas W, and Professional Practice and Guidelines Committee. "Carrier screening for spinal muscular atrophy." Genetics in medicine : official journal of the American College of Medical Genetics vol. 10,11 (2008): 840-2. doi:10.1097/GIM.0b013e318188d069

[47] Ibid.

[48] SMA Genetic Carrier Status Test Being Launched in Europe, Latin America, SMA News Today, April 27, 2017, https://smanewstoday.com/2017/04/27/synlab-and-counsyl-to-launch-sma-genetic-carrier-status-test-in-europe-latin-america/

carrier,[49] it makes sense to provide the test to everyone. In Europe, only Spain, Portugal, and Italy can perform these screenings.[50] In Germany, Austria, Netherlands, and Switzerland, future mothers may ask for a prenatal blood test to determine whether they are carriers.[51] Nevertheless, in this case, the mother is already pregnant, so her choices are more limited regarding the fetus.

Legal Issues

Public policy always needs time to catch up with technology, even when the standard of care is clear. In the case of newborn screening, this standard is evolving. The medical sector cannot only precisely determine the illness, but sometimes actually cure it. There are two parts for the legislation: diagnosis and treatment. Moreover, screening regulations may be complicated because screening before the pregnancy reveals only a possibility of SMA, screening during the pregnancy reveals the presence of SMA, and screening positive for SMA after birth requires treatment. The screening results are not always reliable. For example, if both of the parents are discovered to be SMA carriers during pre-pregnancy screening, they have only a 25% chance having a baby with SMA. Or, for example, when they calculate the possibility during the pregnancy having an infant with Down syndrome based on the blood test of the mother, her age, and the ultrasonography, it is the mother's choice to proceed with amniosynthesis to confirm the condition. Even if these predictions combine with issues of privacy or religious and cultural beliefs,[52] public policy needs to balance the interests around the protection of the health of mother and fetus. But, can public health justify making newborn screening obligatory for the entire population, even if it is a hereditary genetic disease? Or should it be only an option for the parents? Generally, most parents want to know whether their child has a genetic defect and whether there is an available treatment for it. If not, based on cultural reasons, these parents should have an opt-out possibility, where they could refuse these genetic tests. Or, with an opt-in provision, they could choose those tests which they think are appropriate. This kind of regulation would respect the right of an individual's choice as hereditary diseases are not infectious, it is rather a part of private than public health. The states could offer a choice for parents by making newborn screening available for as many diseases as technology allows. Public health agencies promote family planning programs focusing on the health of infants. They inform parents that through newborn screening-detected diseases can be treated with higher efficiency.[53]

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[49] Ibid.

[50] Ibid.

[51] Prenatal Blood Test for SMA Now Available in Four Countries in Europe, BillionToOne Announces, SMA News Today, November 8, 2019, https://smanewstoday.com/2019/11/08/prenatal-blood-test-sma-other-single-gene-diseases-now-available-four-countries-in-europe/

[52] Cooper, Todd: Blood test for newborns faces religious challenge, Omaha World-Herald/December 21, 2004

[53] Kraszewski, J., Burke, T., & Rosenbaum, S. (2006). Legal issues in newborn screening: implications for public health practice and policy. Public health reports (Washington, D.C. : 1974), 121(1), 92–94. https://doi.org/10.1177/003335490612100116

In the United States, newborn screening is mandatory and parental consent is not required. Based on two foundational laws, the states have the responsibility to protect their citizens. The police power protects "the health, safety, morals, and general welfare," the "parens patriae" power (i.e., the state acting as parent in certain circumstances) ensures the rights of children and vulnerable persons to health and well-being. The latter gives the right to the state to intervene against an abusive or negligent parent to protect the child. These state privileges and responsibilities are weighed against parents' rights to make decisions about their minor children's welfare. The parents can accept or refuse medical treatments according to their views.[54]

In the European Union, the establishment of a centralized screening committee would be reasonable as many different aspects can block the decision of whether or not to accept a new screening method in a country. A screening committee would have the necessary expertise to review scientific studies and analyze their advantages and disadvantages. However, a committee for evaluating cost-effectiveness and ethics would also be needed. Although some countries involve parent and patient groups in decisionmaking, in addition to federal and regional health authorities and medical professionals, the final word should be the state's, based on the funding and provisions made by the health care system. Eighteen EU countries already have a committee, laws, and regulations devoted to examining newborn screening. Some of them make it obligatory for parents to have their babies screened. Another group of states only inform them that they have a possibility for screening, and the parents can choose it or not. In the last five years, twenty-one EU member countries have changed the newborn screening policy.

A common mechanism would be desirable between them as national screening committees have to evaluate the same issues. The European Network for Health Technology Assessment (EUnetHTA) asks for collaboration because it can facilitate a transparent and effective health technology assessment.[55]

Public health authorities are responsible for offering newborn screening to their citizens.[56] A legal obligation ensures quality and accessibility. But, it would remain only an offer of screening, and should not be mandatory.[57] In the US and Hungary, newborn screenings are mandatory, under the rationale that parents have the obligation to protect the health of their child. But can a human right become an obligation? At a European Cystic Fibrosis Society's conference, a question was posed and remained unanswered: is

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[54] US Constitution, 14th Amendement

[55] EU Tender "Evaluation of population newborn screening practices for rare disorders in Member States of the European Union", Newborn screening in Europe, Expert Opinion Document, Final 28/08/2011 https://www.isns-neoscreening.org/wp-content/uploads/2018/11/Expert-opinion-document-on-NBS-FINAL.pdf

Kristensen FB, Mäkelä M, Neikter SA, Rehnqvist N, Håheim LL, Mørland B, Milne R, Nielsen CP, Busse R, Lee-Robin SH, Wild C, Espallargues M, Chamova J; European network for Health Technology Assessment (EUnetHTA). European network for health technology assessment, EUnetHTA: planning, development, and implementation of a sustainable European network for health technology assessment. Int J Technol Assess Health Care. 2009 Dec;25 Suppl 2:107-16. doi: 10.1017/S0266462309990754. PMID: 20030898.

[56] Charter of fundamental rights of the European Union, Art 24

[57] CoE additional Protocol, art 10

newborn screening a basic human right?[58] They based this theory on the concept that if doctors detect this genetic illness less than two months after birth, the child has better chances of survival. The same can be applied to SMA, but no one asked this question yet. It is an important question now that Zolgensma is accessible and should be given before the baby's second birthday.

To have a look at it from a broader perspective, is health care a basic human right? If so, then providing it is a duty of the state. From a narrower perspective, is early detection of a lethal genetic disease a basic human right? After several debates and studies, we can answer "yes" to this question if four conditions are present: 1) if the child is born in a region where the questioned disease is relatively common (at least 1:10,000 in the case of cystic fibrosis); 2) the newborn screening program exists there or can be created; 3) in case of a positive test, there is an available treatment; and 4) sustained funding is available. We must answer "no" to the question above if it causes more harm than good, when the regional readiness does not exist or when there is no available funding, service, or treatment.[59] Taking into consideration, that we have talked about newborn screening programs in developed countries above, of whom all of the four points are true, making newborn screening available for every parent would be a duty.

Summary

Prevention should always come first before treatment. It would be beneficial for the state as they would have less material expenditure for life aids, hospitalization, therapies, and medication for the patients. Paying for Spinraza through health insurance is expensive. Moreover, the mother of a sick child probably cannot continue her career because she will most likely need to stay at home to care for her son/daughter. It is a loss for the individual as she did not have a choice and also a loss for society regarding the labor market. Among the goals of the World Health Organization (WHO), we can find inter alia the prevention of non-communicable diseases, the promotion of mental health, the improvement of access to essential medicines and health products, and the improvement of monitoring, data, and information.[60] In this way, they could urge the development of newborn screening, provide mental support for the patients' families, negotiate the price of life-saving drugs, promote the invention of new treatments and methodize the registration of genetic disease. But there is also work to be done at the national level. For example, the Minister of Health in each country could focus more on the importance of the Guthrie test and increase the revenue sources for health services. Developed countries should find the financial sources to offer all technologically available newborn tests to the people. It is the state's responsibility to protect its citizens and this test can save lives. Making SMA screening available to parents as part of standard newborn screening ensures a basic human right.

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[58] Farrell PM. Is newborn screening for cystic fibrosis a basic human right? J Cyst Fibros. 2008 May;7(3):262-5. doi: 10.1016/j.jcf.2008.01.001. Epub 2008 Feb 11. PMID: 18262856; PMCID: PMC2504861.

[59] Ibid.

[60] What we do, WHO https://www.who.int/about/what-we-do