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Are laws the solution for rare diseases?

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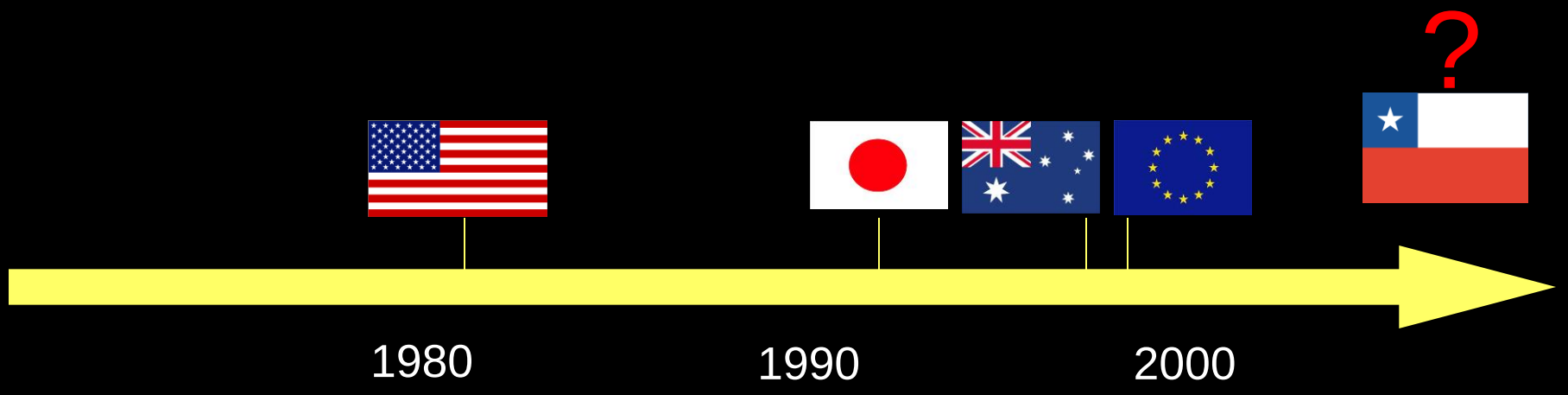
- Unpaid activities: Member of the Board of Directors of “Doctors Without Brand”, Law 20.850 Citizen Surveillance Commission, Member of the Ministerial Commission for RD, Former President of the Latin American Society of IEM and Newborn Screening (2013-2015), LatAM representative in International Society for Neonatal Screening (ISNS).

Orphan drugs laws

•They establish incentives for the development of drugs for rare diseases.

- Oncology and Genetic/IEM >70%



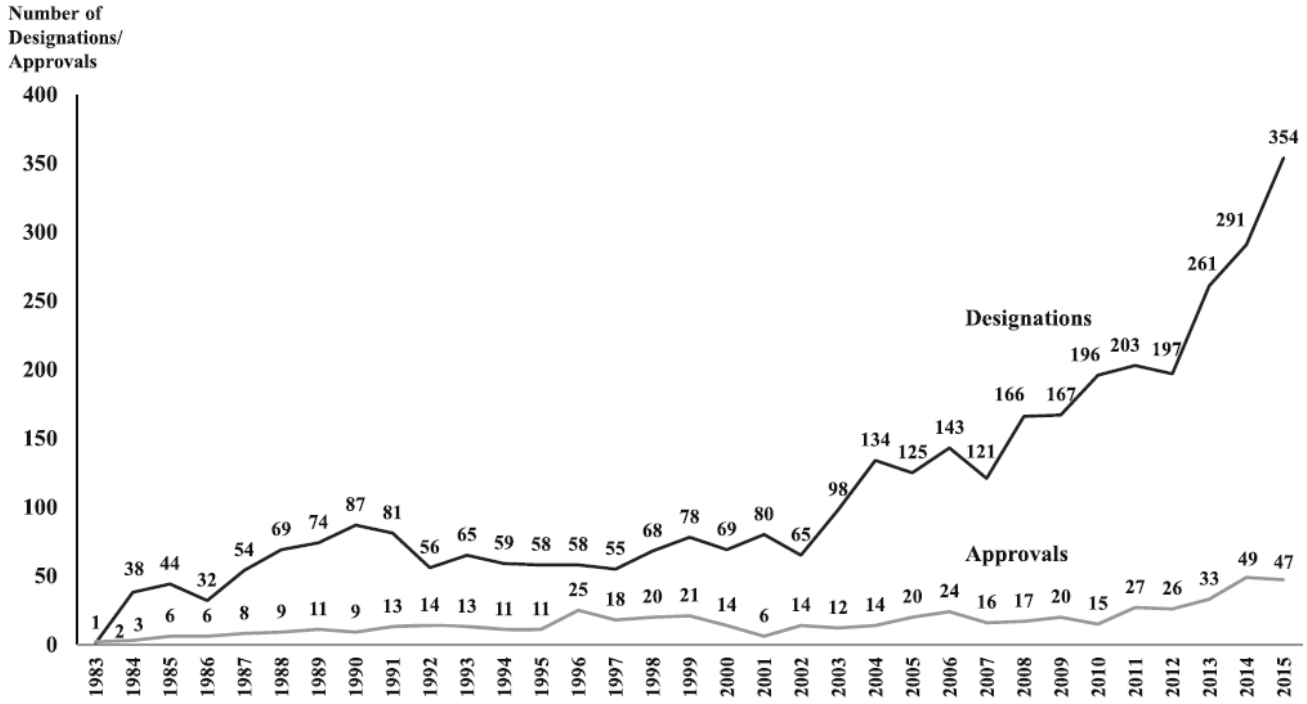


Fig. 1 FDA Orphan Drug Designations and Approvals, 1983–2015

Liberté • Égalité • Fraternité
RÉPUBLIQUE FRANÇAISE

Ministère de la santé
et de la protection sociale

Secrétariat d'Etat
aux personnes handicapées

ministère
éducation
nationale
enseignement
supérieur
recherche

ministère délégué
à la recherche

French National Plan for Rare Diseases
2005 – 2008

"Ensuring equity in the access to diagnosis, treatment and provision of care"

Coverage regulations for RD in Chile

- Law 18.469, 1985: Creates a general health benefits regime: general public health programs, preventive medicine examination, pregnancy care and comprehensive curative medical assistance: consultation, examinations, procedures, hospitalization, treatments and medications from the National Formulary.

- Law 19.966, 2004: Within the previous regime, it creates a general guarantee regime through which it generates coverage obligations for the National Health Fund. It also creates the explicit guarantee system (GES), applicable to FONASA and ISAPREs.

- Law 20.850, 2015: Creates within the RGG, a specific system for high-cost diagnoses and treatments.

- Law 21.258, 2020: In a way not linked to the previous systems, it generates a fund for various activities related to cancer, among which are some related to therapeutic benefits, including drugs.

Thanks Dr Dino Sepúlveda. MINSAL

Coverage and access of RD in Chile

- System of Explicit Health Guarantees (2006): Cystic Fibrosis, Hemophilia, Cancer in boys and girls.

- Law 20.850 (2015): Mucopolysaccharidosis I, II and VI Tyrosinemia type I, Gaucher disease, Fabry disease, Hereditary angioedema, Pancreatic neuroendocrine tumors, Generalized dystonia, Primary immunodeficiencies, Huntington's disease, Epidermolysis bullosa, Amyotrophic lateral sclerosis, Severe hypoacusis, Unresectable stromal GI.

- Newborn screening for PKU and CTH (1992).

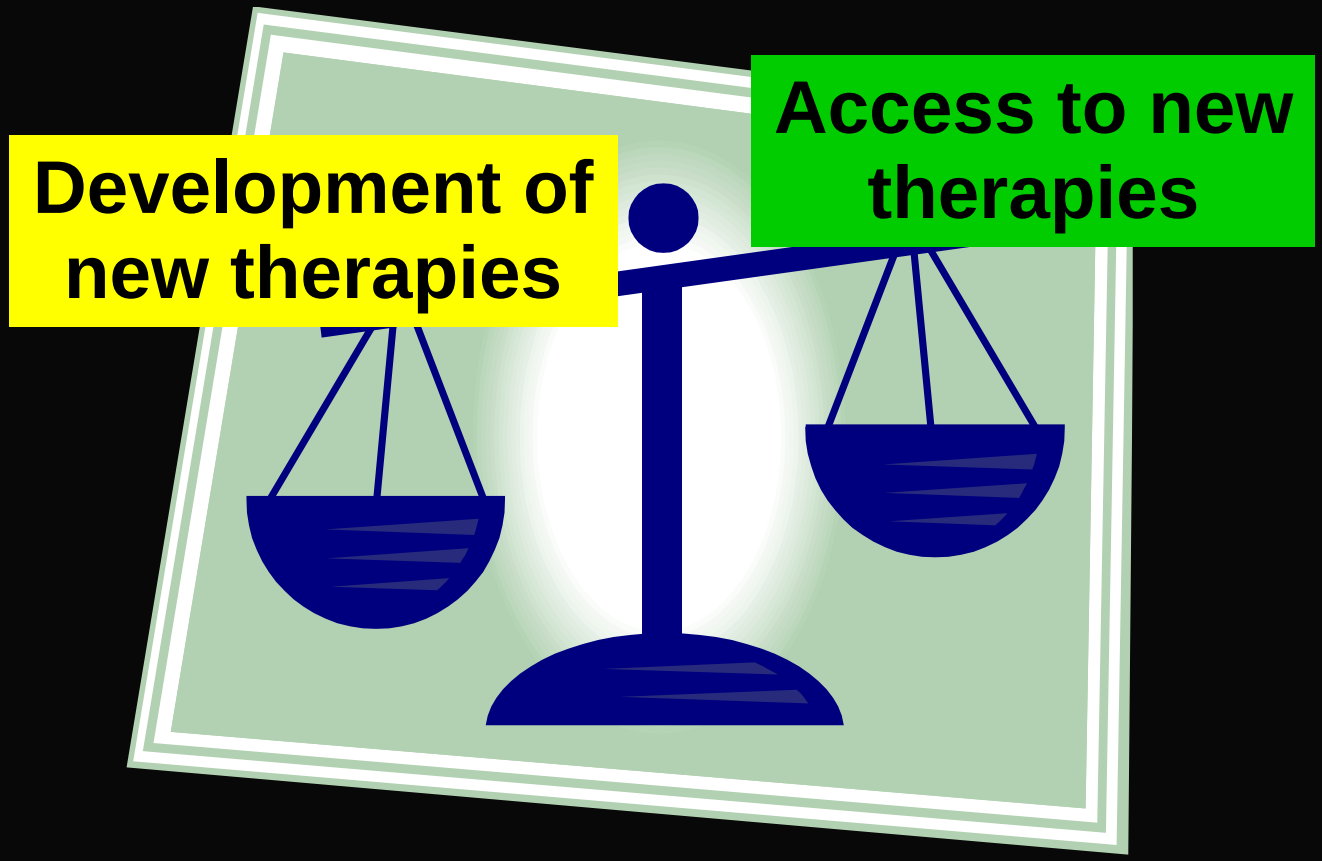
Thanks Dr. Dino Sepúlveda. MINSAL

- The Universal Declaration of Human Rights, proclaimed by the United Nations General Assembly in 1948, establishes the right to life, ratified in the European Convention on Human Rights in 1950.

- The right to the "highest attainable standard of health" was first proposed in 1946 in the WHO constitution. Since then, the right to health has been incorporated into different international treaties and declarations and constitutionally protected in most developed countries, with the notable exception of the US.

- The European Court of Human Rights (ECHR) has interpreted the right to life as the protection of medical care.

European Court of Human Rights: The European Convention. 1948. http:// www.echr.coe.int/pages/home.aspx?p=basictexts. Accessed 21 Mar 2016.
World Health Organization. Constitution of the World Health Organization. 1946.
Gericke CA, Riesberg A, Busse R. Ethical issues in funding orphan drug research and development. J Med Ethics. 2005;31(3):164–8.



Rodriguez-Monguio et al. Ethical imperatives of timely access to orphan drugs: is possible to reconcile economic incentives and patients' health needs?
Orphanet Journal of Rare Diseases (2017) 12:1

fairness requires “a positive action by the state [or government] when the market does not provide a good match between investments and health [care] needs. Finally, fairness requires that the barriers to access should be morally justifiable”

Rhee TG.
Policymaking for Orphan Drugs and Its Challenges.
AMA J Ethics. 2015 Aug 1;17(8):776-9.

SOLUTIONS

- Modifications to regulations in drug-producing countries (orphan drug act).
- Generation of legislation that comprehensively addresses the problem of high-cost drugs (access, patient participation in clinical studies, education).
- Diagnostics and production of "non-profit" drugs.
- Regulation of conflicts of interest.

NATIONAL PLAN PROPOSAL

FOR RARE DISEASES

WORK METHODOLOGY

The work was structured in 5 commissions that addressed the strategic lines of the plan:

a. Steering, Regulation and Supervision Commission.
b. Commission for the Provision of Assistance Services.
c. Education, Promotion and Training Commission.
d. Registration, Surveillance and Information Commission.
e. Commission for Protection and Social Insertion.

Each commission was made up of:

- **Commission Coordinator (Senator Goic Parliamentary Office representative).**
- **Executive Secretary, democratically elected by the participants of the Commission.**
- **Participants: people with RD, their relatives, caregivers; healthcare professionals; academics, industry representatives.**

The commissions met weekly. A minute of each meeting was drawn up.
They worked on a report form for each commission structured based on the essential elements of the proposed plan.

NEXT STEPS

- Institutionality: National Commission of RD with patient participation.
- Incorporation to Orphanet (information, visibility, education, registry, etc).
- Incorporation of RD to Digital Health (tele dysmorphology, RD telecommittees).
- Expansion of the NBS from 2 to 26 conditions, national and universal coverage.

Are laws the solution for rare diseases?

“We need a set of regulations adapted to each country / region, considering previous history, current situation and future challenges.”