

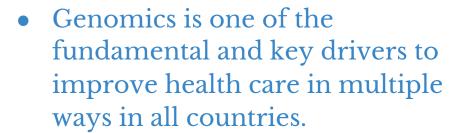


Education and workforce

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 Health systems strengthening should be taken into account, such as education and training of healthcare professionals.



Priority issues about genomics

Clinical Genetics

Professionals

33 active clinical geneticists (1/500,000 inhabitants)

1 training program (Hospital Clínico Universidad de Chile)

Areas

Pediatrics, Neonatology, Maternal-fetal Medicine, Obstetrics and Gynecology, Internal Medicine, Hereditary Cancer

Clinical laboratories

15 clinical cytogenetics laboratories (caryotype, FISH)

3 molecular diagnosis labs (MLPA, chromosome microarrays, Sanger sequencing for limited number of genetic dideases)

6 molecular pathology laboratories



Training program in Clinical Genetics

- Duration of three years
- Four modules: scientific method, biological and molecular basis of genetics, laboratory genetics and clinical genetics
- Rotations through all genetic services in Chile
- Rate of new graduates is 0.7 per year
- Residents from other specialties: Obstetrics and Gynecology, Maternofetal Medicine, Endocrinology, Hemato-oncology, Neurology



Telemedicine: Telegenetics

- The majority of clinical geneticists in Chile work in the Metropolitan Region (78%).
- The second cause of infant mortality (7 / 1,000 live births) in Chile is congenital anomalies with 35%.
- Since December 2015 there is a mandatory National Registry of Congenital Anomalies in Chile, to annotate and describe anomalies in all newborns.
- Telemedicine for assistance and educational services at regions
- Support to the Ministry of Health and its health network
- Integrate primary care centers with hospitals and university centers.



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Telemedicine: Telegenetics

• Perinatal Telemedicine Program of **CERPO** (Oriente Perinatal Reference Center), available to Health Services that refer pregnant women whose fetuses have congenital malformations, live and with remote sonographic examinations. Regional contacts: Hospitals of Antofagasta, Easter Island, Temuco, Concepción, Valdivia, Osorno, Coyhaigue and Punta Arenas.

 Establish a protocol for remote consultations in clinical genetics of hospitalized or outpatients to optimize the care, advise and train health professionals and improve access to medical specialists in genetics; for clinical cases with suspicion of diseases of genetic origin.





Roadmap for the development of Genetics and Genomics in Chile

Sociedad de Genética de Chile to Ministry of Health:

Provision of health services

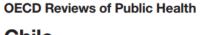
Generation of new knowledge (Chile spends 0.36% of GDP in research (average for OECD countries is 2.34%)

Public education and awareness

Regulation

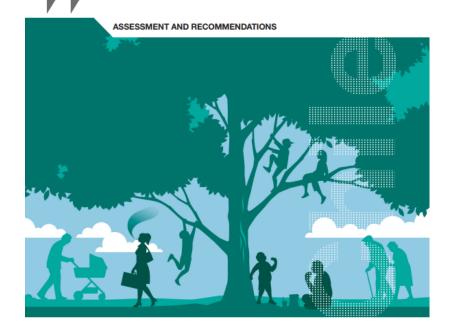


International collaboration





A HEALTHIER TOMORROW







Policy recommendations for improving public health in Chile: use genomics to strengthen preventive care:

- Develop a national precision medicine strategy that sets out equitable access to genomic medicine, safeguards against genetic discrimination, workforce development and investment in basic research;
- Develop a framework for personal genetic data, that enables data sharing for precision medicine services and research, whilst protecting individual privacy;
- Establish quality assurance which guarantees standards across laboratories undertaking genetic analysis;
- Increase "genetic literacy" amongst health professionals and the public.

