

Genetic service provision in Argentina

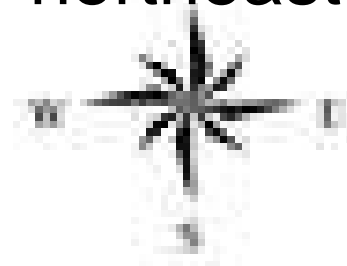
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A map of South America with Argentina highlighted in blue. The word "Argentina" is written in large, bold, black letters across the top of the highlighted area.

Argentina

- Is a South-American country, fourth in size in the continent after Canada, United States and Brazil and eighth in the world. Argentina occupies a continental surface area of 2,766,890 km² between the Andes mountain range in the west and the southern Atlantic Ocean in the east and south.
- It is bordered by Paraguay and Bolivia in the north, Brazil and Uruguay in the northeast, and Chile in the west and south.



Economic Crisis and Recovery

- By 2002, Argentina had defaulted on its USD 150 billion foreign debt, its GDP had shrunk, unemployment was more than 25%.
- However, careful spending control and heavy taxes on now-soaring exports gave the state the tools to regain resources and conduct monetary policy, leading to a significant economic recovery.

Economic Crisis and Recovery

- By 2006, the situation further improved. The economy grew 8.8% in 2003, 9.0% in 2004, 9.2% in 2005, and in 2006 it was on the same track though. However, inflation, estimated at around 10 to 12%, has become an issue again, and income distribution is still considerably unequal.

Demographic and Health Indicators

- In 2001, the population of Argentina was about 36 million (third in South America in total population).
- Argentina's population density is 14 inhabitants per square kilometer. However, the population is not evenly distributed: areas of the city of Buenos Aires have a population density of over 14,000 inhab./km², while Santa Cruz province has less than 1 inhab./km². Argentina is the only nation in South America with a net positive migration rate of about +0.4 persons.

ARGENTINA

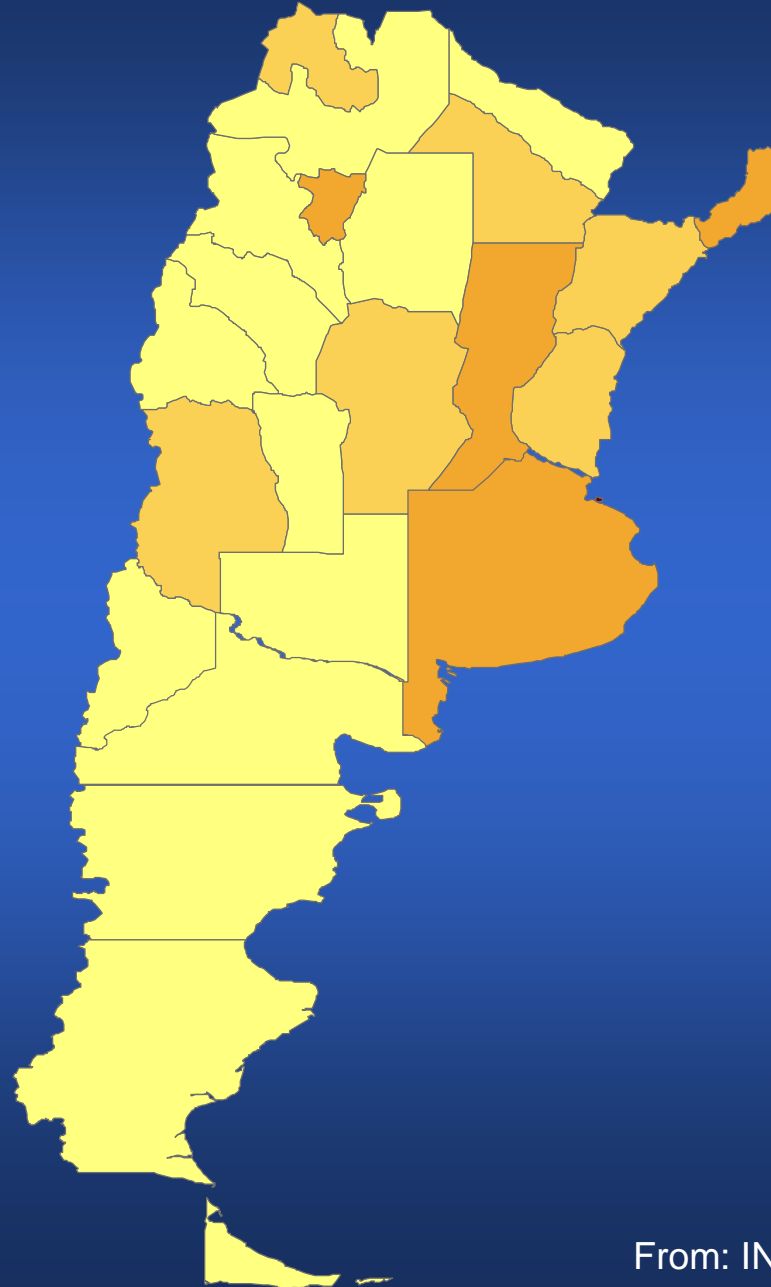
Population Density

Population: 37.869.723
Area: 2,7 million sq km

Country Population
density: 13.8 inhabitants
per sq km

Population Density

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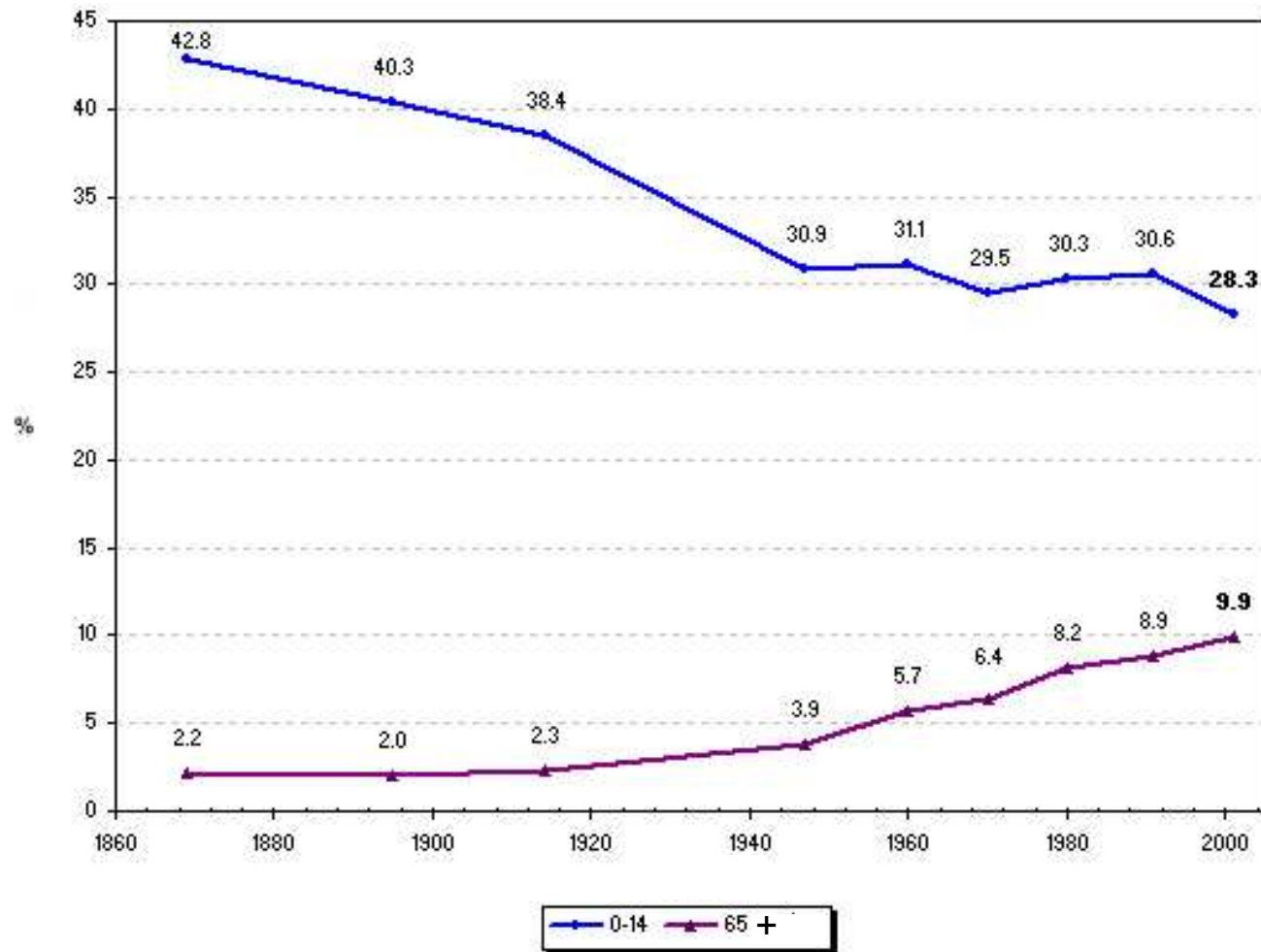
From: INDEC, 2003

Demographic Profile

- Argentina's population is highly urbanized. About 3 million people live in the city of Buenos Aires, and 9 million in the metropolitan area around the city (Greater Buenos Aires), making it one of the largest urban conglomerates in the world.
- The second- and third-largest cities in Argentina, Cordoba and Rosario, comprise about 1.3 and 1.1 million inhabitants respectively.

	Total	Buenos Aires
Annual no. of births (thousands)	698	42
Population under 15 (percentage)	22	18
Infant mortality rate (per thousand live births)	13.3	9.6
Gross birth rate (thousand)	18.2	13.9
Gross mortality rate (thousand)	7.6	11.1
Mean Annual growth rate (1991-2001) (per thousand persons)	10.1	-6.3
Total fertility rate (per woman)	2.30	1.39
Life expectancy at birth in years:	71.93, 68.44 (male) 75.59 (female)	72.72 69.17 (male) 76.45 (female)
Population aged 65+: (percentage),	9.94	17.05

Population (%) under 15 and over age 65 (1869-2001)



..... INDEC. Population census.

ARGENTINA

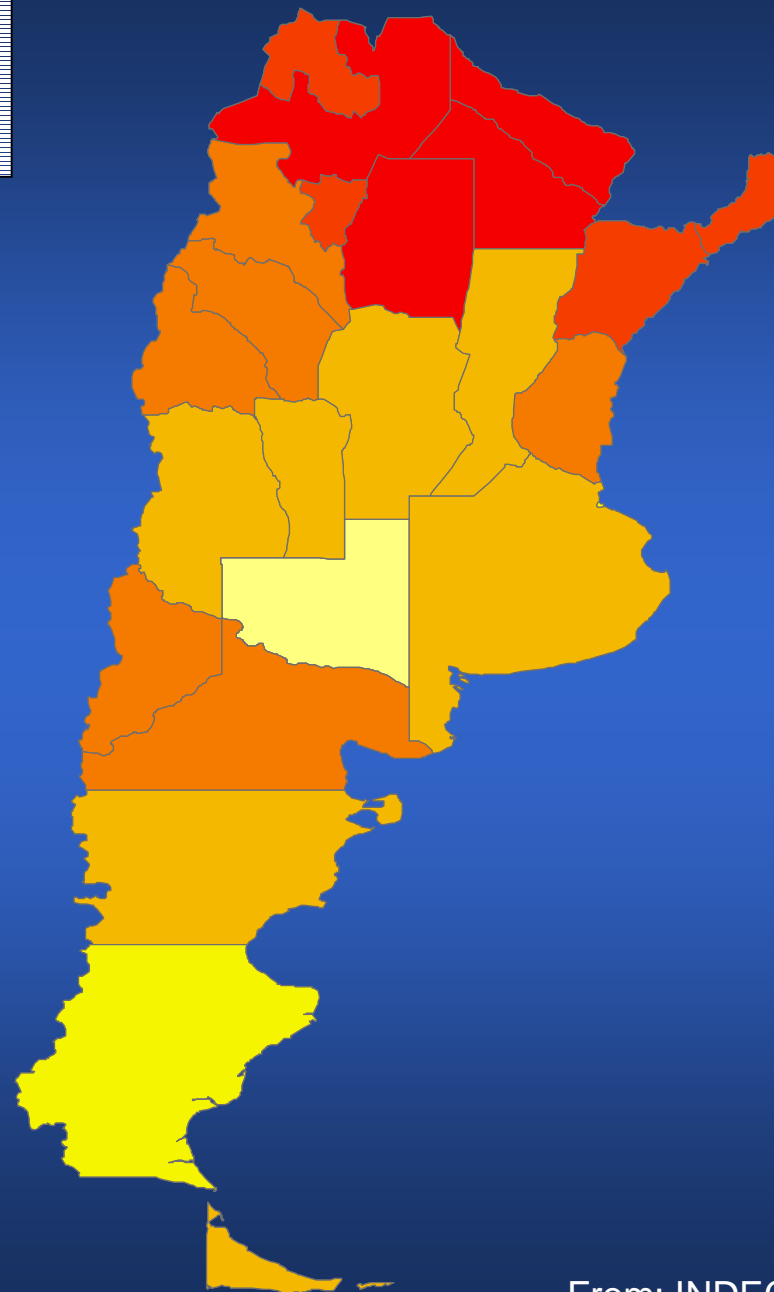
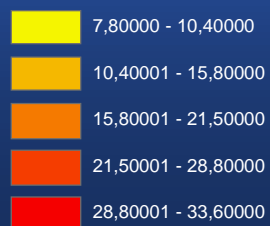
Unsatisfied basic needs

(Percent)

Country: 17.7%
Ciudad Buenos Aires:
7.8%

NBI Percent

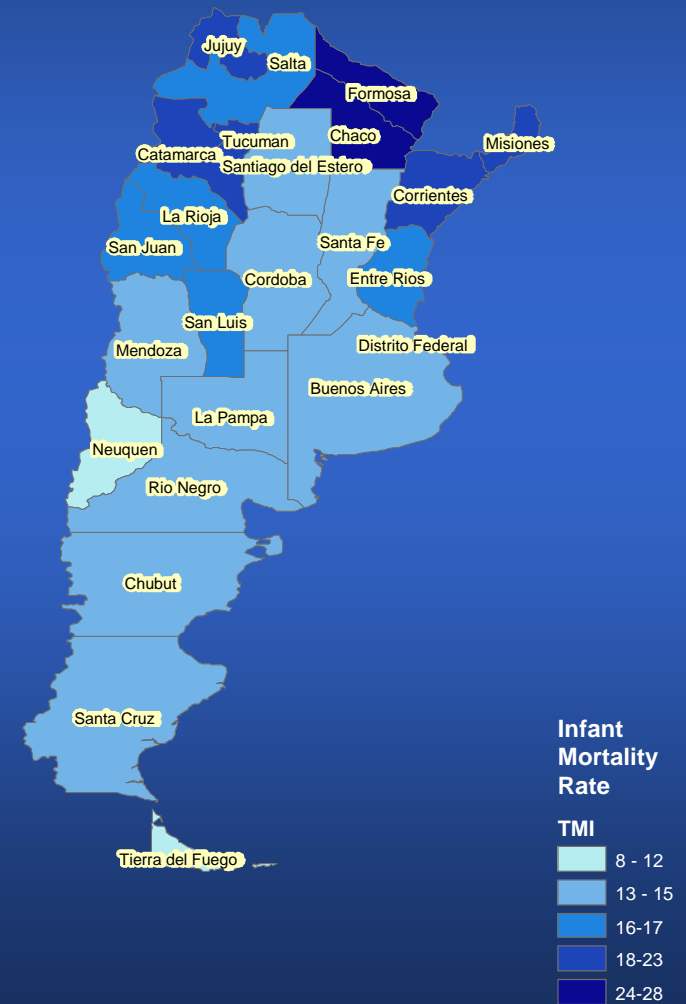
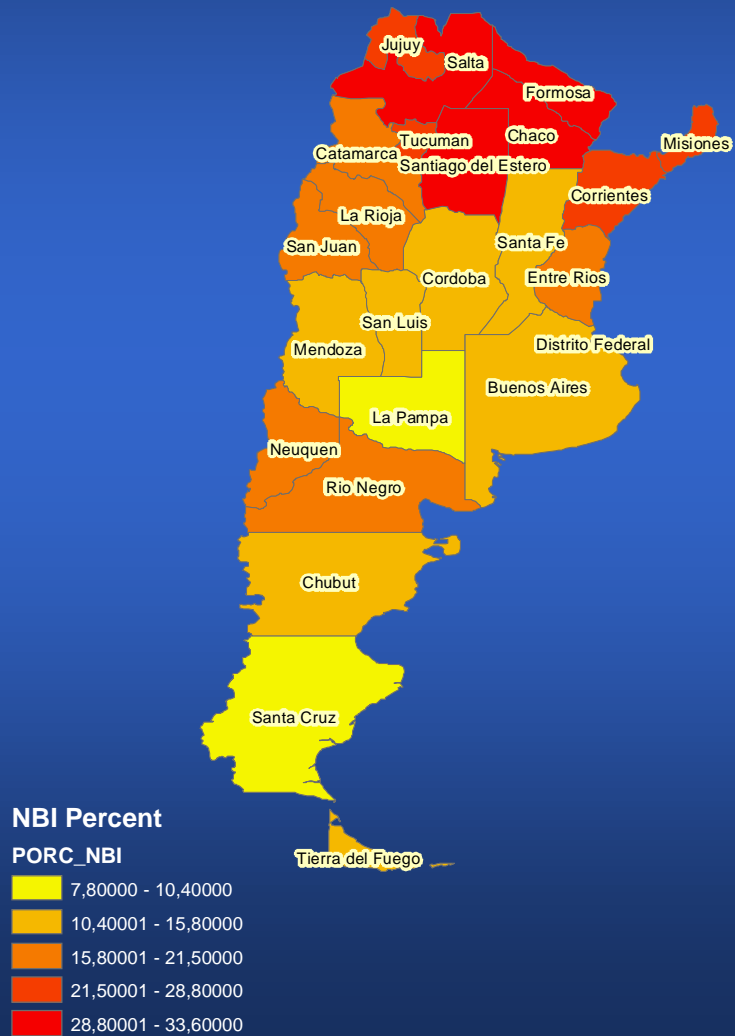
PORC_NBI



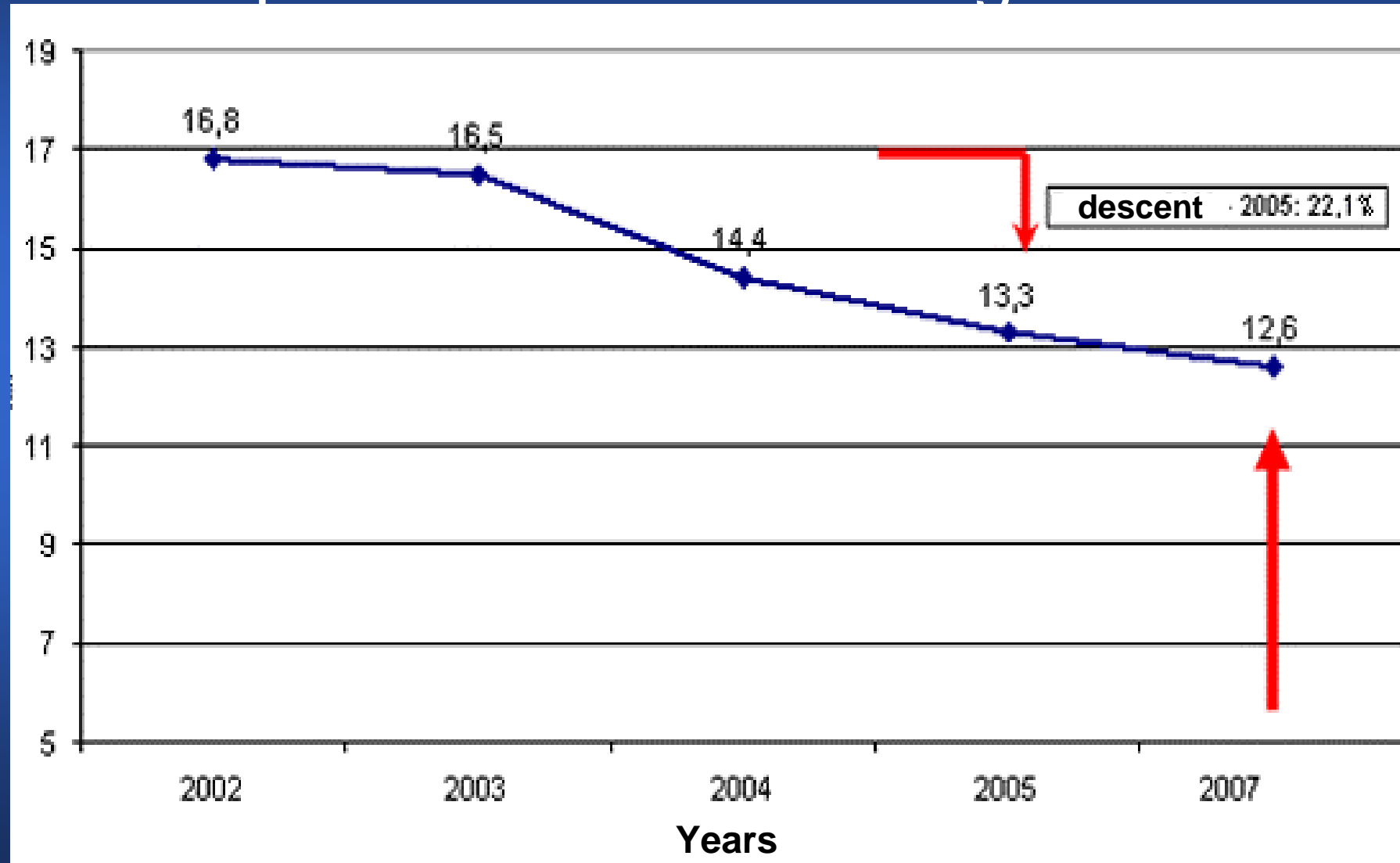
From: INDEC, 2001

Relationship between Unsatisfied Basic Needs and Infant Mortality

$r = 0,71$



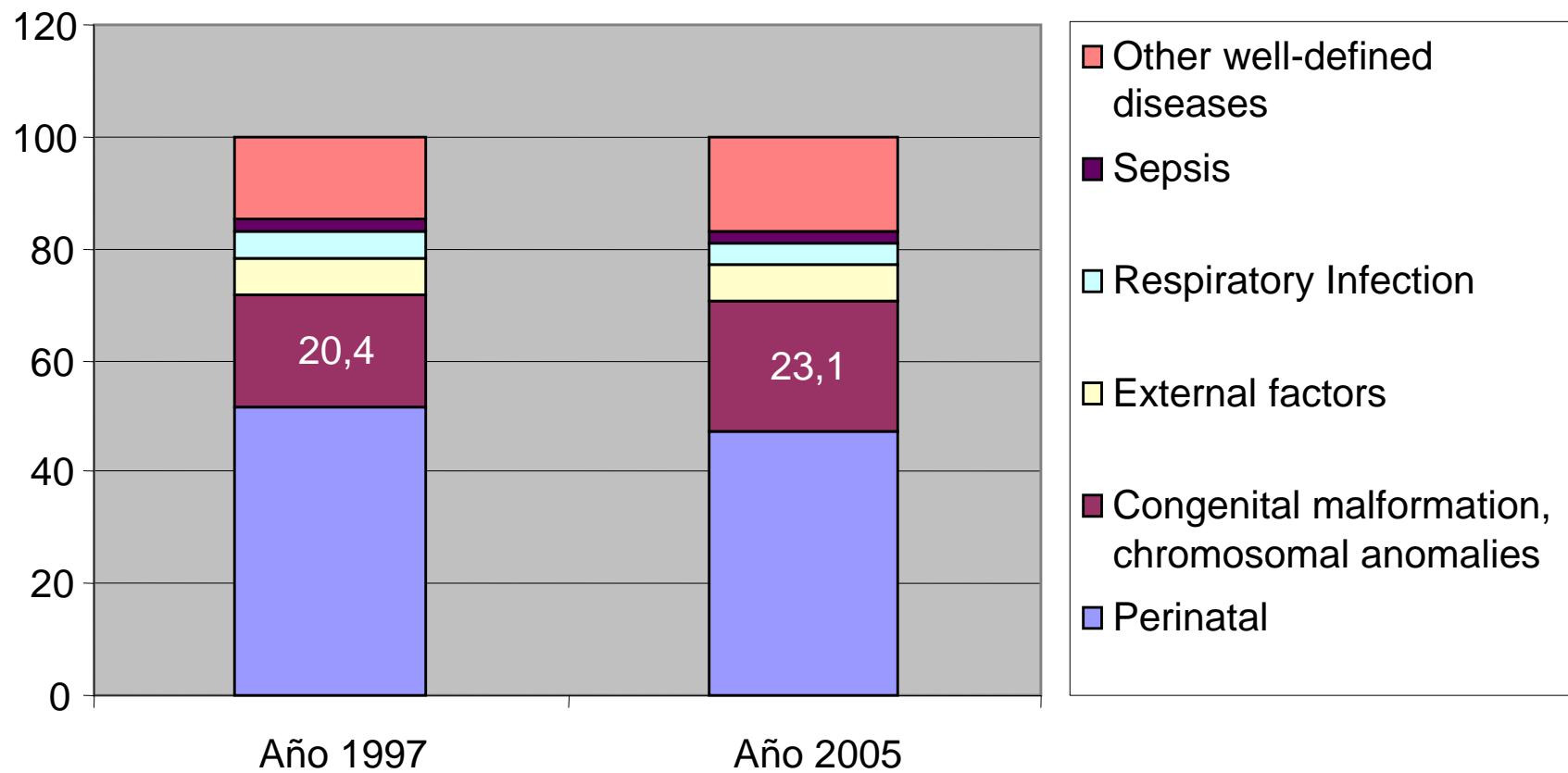
Infant Mortality Rate 2002- 2005 and expectation for 2007. Argentina



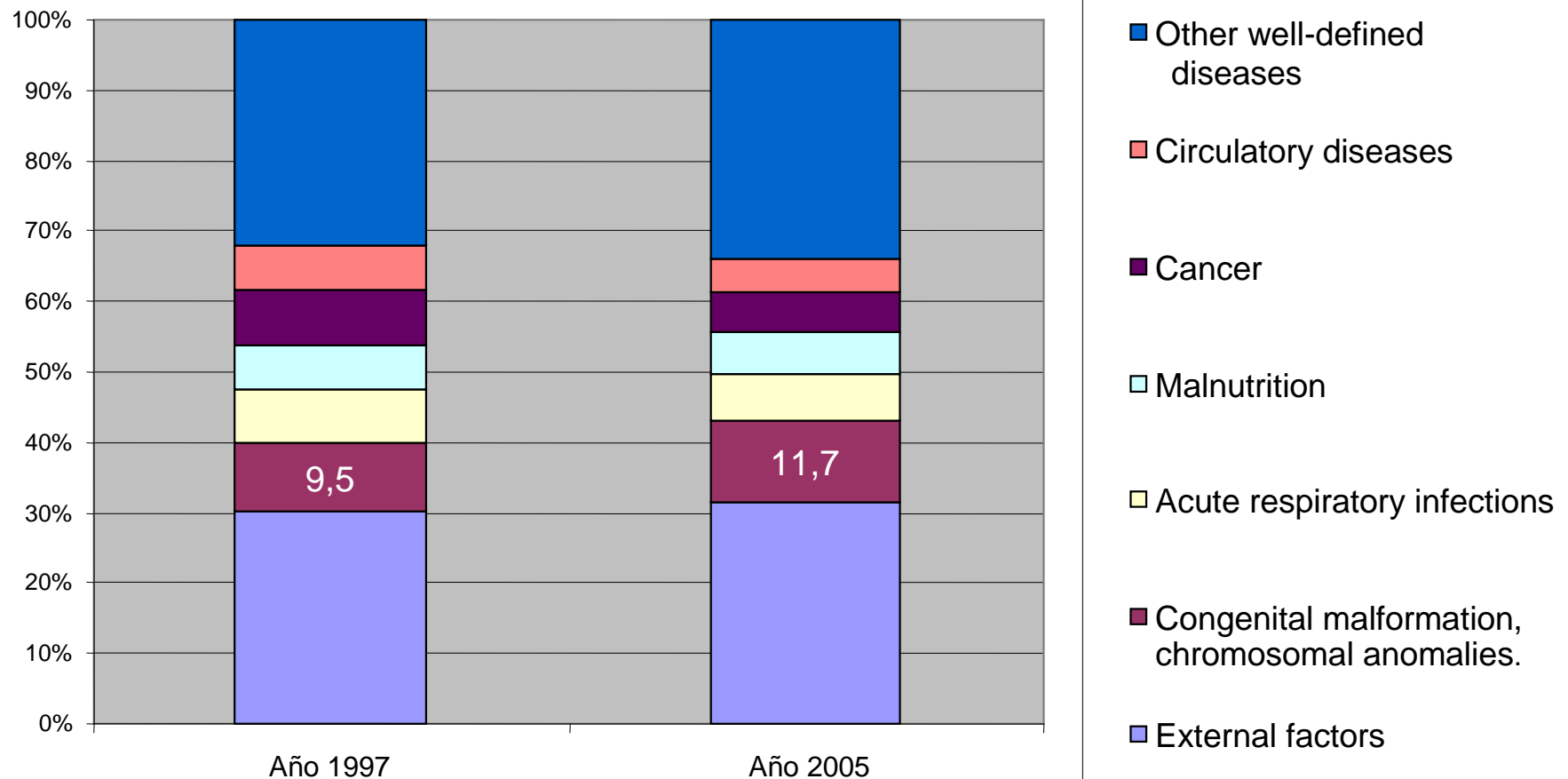
INFANTS DEATHS DUE TO CONGENITAL MALFORMATIONS

- In 1997 and 2005, the absolute number of infant deaths due to congenital malformations were 2,475 and 2,403 respectively, but their relative contribution to infant mortality increased from 20.4 % to 23.1 %, as a consequence of the decrease in infectious and nutritional diseases.

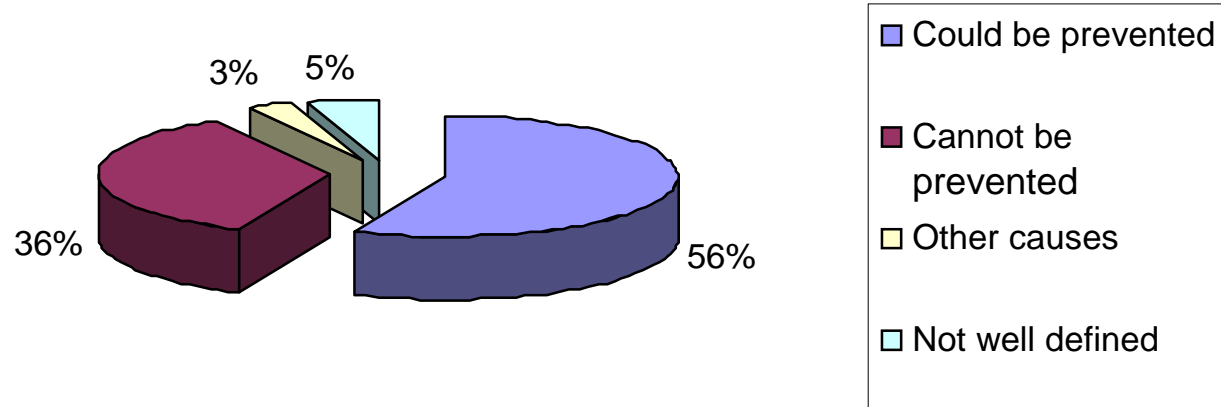
MAIN WELL-DEFINED CAUSES OF DEATH IN CHILDREN in the FIRST YEAR OF LIFE



MAIN WELL-DEFINED CAUSES OF DEATH IN CHILDREN BETWEEN 1 AND 4 YEARS OLD. Argentina, 1997-2005.



Total infant deaths 2004



Per capita expenditure on health, 2003

			2003	
Per capita total expenditure on health (US\$)			Arg 305	USA 5711
Per capita government expenditure on health(US\$)			148	2548
General government expenditure on health as % of total expenditure on health			48.6	44.6

Registry of Congenital Malformations

- ECLAMC, the Latin-American Collaborative Study on Congenital Malformations is active in Argentina. It is a surveillance program for the detection of congenital defects, which is carried out at maternity hospitals in several Latin-American countries. It's figures show that the prevalence at birth of congenital malformations is similar to that of developed countries

History of Medical Genetics in Argentina

- In 1969 the Medical Genetics Centre of the Ministry of Health was founded for research, educational, and medical care purposes.
- In 1963 and 1971 Genetics Departments were created at the two then existing Pediatric Hospitals in Buenos Aires
- In 1988 the Department of Genetics started working at the newly created Garrahan Pediatric Hospital, which is now the main tertiary referral centre in the country.

Current State of Departments of Genetics in Buenos Aires

- In 1991, the Ministry of Public Health recognized Medical Genetics as a medical speciality. Currently there are approximately 100 clinical geneticists.
- Genetic services are concentrated in the city of Buenos Aires, which counts with genetic departments in several public hospitals (three pediatric hospitals, the university hospital and the Center of Medical Genetics of the Ministry), employing approximately 20 clinical geneticists and 30 laboratory geneticists. In addition, the City Health Department funds a clinical geneticist to provide genetic consultations in some of the remaining city hospitals.

Medical Genetics in Buenos Aires

- One of the two main maternity hospitals in the city provides diagnostic services in newborns with congenital malformations, including cytogenetics.
- Several private non profit medical institutions also provide comprehensive genetic services.
- Prenatal diagnosis is, almost without exception, not provided in the public sector (while in private it is thriving).

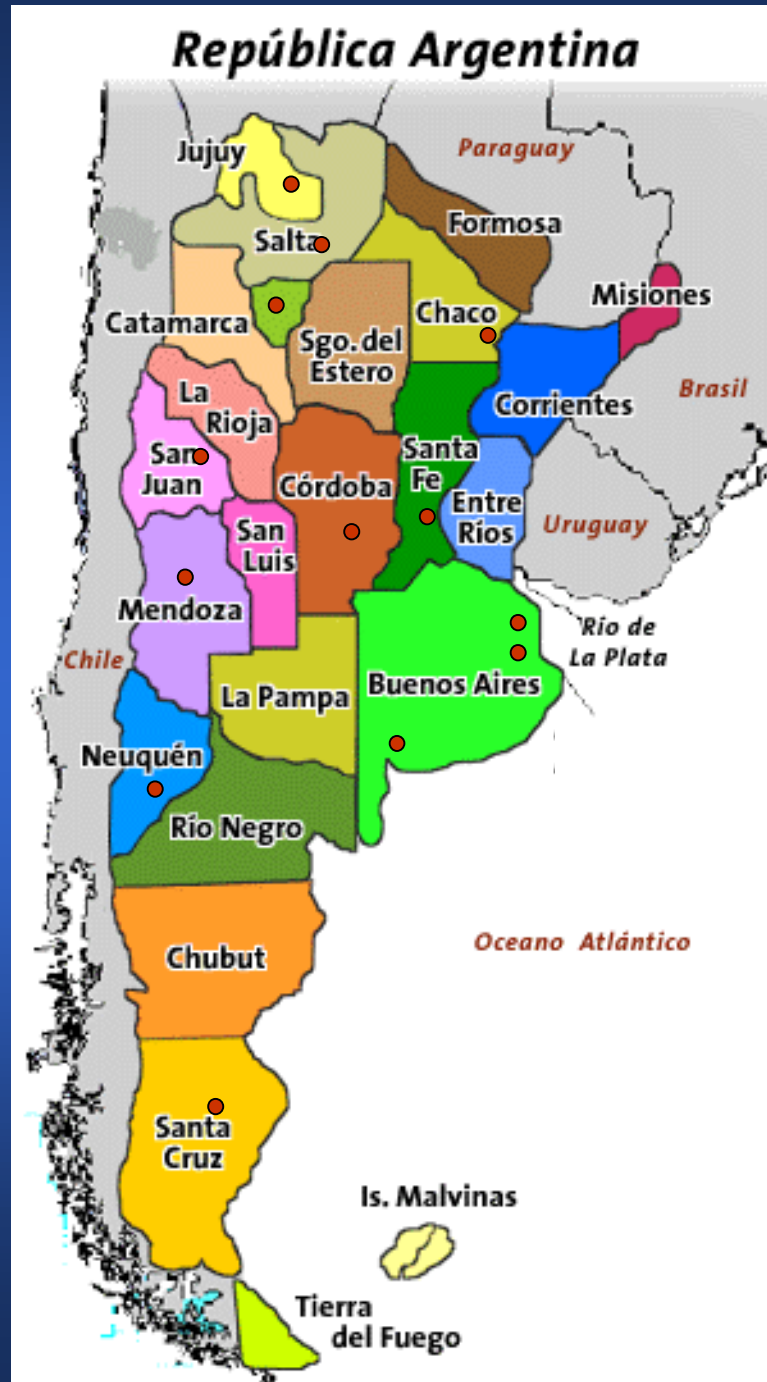
Medical Genetics in the Provinces

The largest cities in Argentina (Rosario, La Plata, Cordoba, and Mendoza), count with clinical geneticists, mostly in large hospitals, and cytogenetic and DNA tests are performed.

However, the development of genetics is deficient in smaller cities, where geneticists lack not only the means to study patients but also the opportunities to improve and update.

Genetic Centres in Argentina

(red dots)



Newborn Screening in Buenos Aires (90% coverage)



- Congenital hypothyroidism
- Phenylketonuria
- Cystic Fibrosis
- Congenital Adrenal Hyperplasia
- Galactosemia.
- Biotinidase Deficiency
- Chagas' disease
- Syphilis
- Congenital Hearing Loss

Some Problems

- Economic difficulties which affect the country are a hindrance to progress in the field.
- Until recently, government officials were not considering genetic services as a priority.
- Therefore, the development of clinical genetics as well as the access of the population to specialized care has been deficient.

Confronting the Problems

- Need for training of more professionals in the different genetic sub-disciplines and the creation of hospital positions in the neglected areas of the country.
- Development of a regional network of genetic services, with interconnected levels of complexity and roots at the primary health care level

Recent Actions of the Ministry of Health

- Creation of a National Program of Community Physicians for Primary Health Care (genetic training is pending)
- Creation of a National Commission of Genetics and Health
- Implementation of a needs assessment of genetic services based on a national survey conducted in 2006, to be followed soon by:
 - creation of new clinical positions and laboratory equipment and personnel
 - implementation of a regionalized network of genetic services



About Genetic Defects

You can

Know clinical signs and natural history of the most frequent genetic conditions for a multidisciplinary follow up.

✓ Give basic genetic information to patient and family specially in multifactorial defects such as congenital heart disease, cleft lip/palate, and neural tube defects.

✓ Coordinate attention of patients with complex genetic pathologies.

✓ Be aware of the psychological and social characteristics of families with one or more members affected with or susceptible to a genetic disease.

✓ Know how to have access to genetic studies that are of benefit to the patient.

✓ Know how to refer patients that need secondary or tertiary genetic care.

Refer your patient to the respective Regional Hospital where the Department of Genetics may resolve your problem.

In more complex cases, get in touch with a tertiary care centre.

You must

Identify patients that may benefit from genetic counselling:

1) Those who have a genetic disease.

Patients who present with:

✓ Two or more major malformations or one major and several minor malformations.

✓ Mental retardation without a known cause, specially when associated with malformations or a positive family history.

✓ Neonatal hypotonia, coma, or acidosis of unknown cause.

✓ Progressive neurological deterioration.

✓ Ambiguous genitalia.

✓ Pathological short stature.

✓ Underdeveloped secondary sexual characteristics.

✓ A phenotype compatible with a known genetic syndrome.

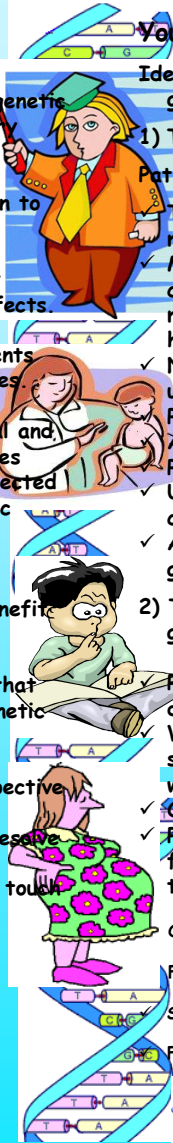
2) Those who are at risk of transmitting genetic disease:

✓ Parents who are gene carriers of diseases of clear Mendelian inheritance.

✓ Women with antecedents of repeated spontaneous abortions, stillborn infants without a diagnosis, or infertility.

✓ Older mothers.

✓ Pregnant women in whom anomalies were found on ultrasound or in laboratory tests.



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