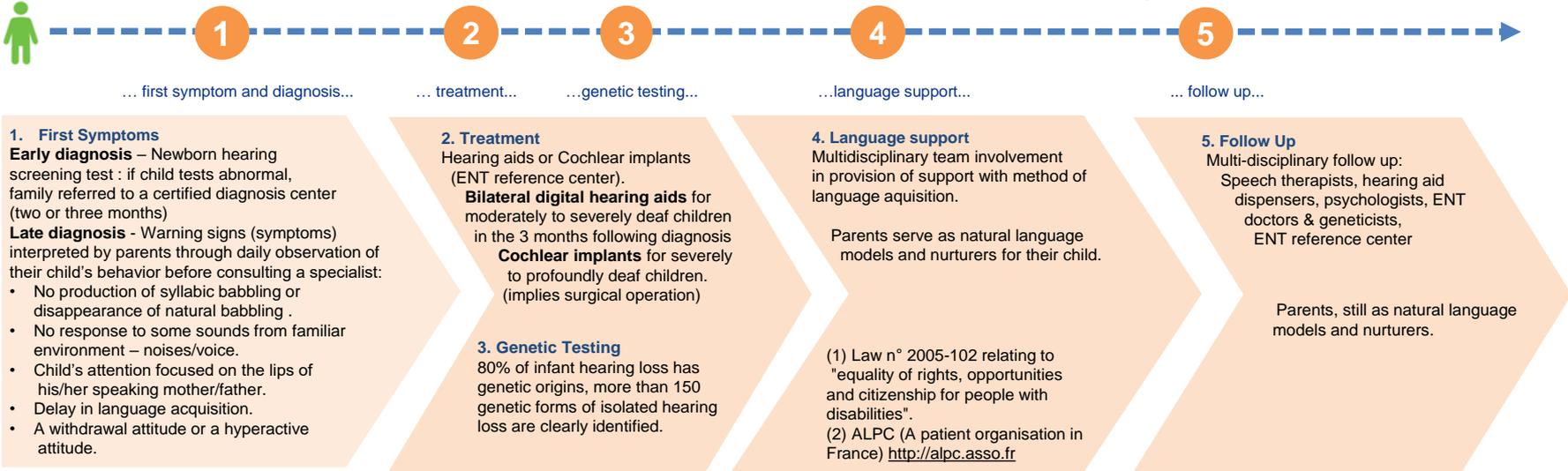


# A patient journey of a patient with Genetic Hearing Loss\*

This is a working document.  
Last updated: July 2021

>90% deaf children born to hearing parents who have no knowledge of deafness or how to deal with it.

Deaf people may not consider their hearing loss as a disease but as a difference and part of their identity



**1. First Symptoms**  
**Early diagnosis** – Newborn hearing screening test : if child tests abnormal, family referred to a certified diagnosis center (two or three months)  
**Late diagnosis** - Warning signs (symptoms) interpreted by parents through daily observation of their child's behavior before consulting a specialist:

- No production of syllabic babbling or disappearance of natural babbling .
- No response to some sounds from familiar environment – noises/voice.
- Child's attention focused on the lips of his/her speaking mother/father.
- Delay in language acquisition.
- A withdrawal attitude or a hyperactive attitude.

**Needs:** Inform the parents on newborn hearing screening test during pregnancy. Convey a positive vision of the child's future and abilities.

Prevent the risk, for the child, of not developing an efficient means of communication in infancy

**Ideally:**  
Early diagnosis

**2. Treatment**  
 Hearing aids or Cochlear implants (ENT reference center).  
**Bilateral digital hearing aids** for moderately to severely deaf children in the 3 months following diagnosis  
**Cochlear implants** for severely to profoundly deaf children. (implies surgical operation)

**3. Genetic Testing**  
 80% of infant hearing loss has genetic origins, more than 150 genetic forms of isolated hearing loss are clearly identified.

**Treatment Needs:** Stimulation of auditory functions.

**Ideally:** Auditory training using the optimum brain plasticity of early age. Stimulating the hearing function creates the brain connections necessary for developing further skills in a spoken language.

**Genetic Testing Needs:** Finding out about

- the mode of inheritance.
- possible associated syndromic symptoms.
- the possible evolution of deafness.

**Ideally:**  
Setting up new clinical trials and giving access to efficient medical care.

**4. Language support**  
 Multidisciplinary team involvement in provision of support with method of language acquisition.  
 Parents serve as natural language models and nurturers for their child.

(1) Law n° 2005-102 relating to "equality of rights, opportunities and citizenship for people with disabilities".  
 (2) ALPC (A patient organisation in France) <http://alpc.asso.fr>

**Needs:** Parents need for an independent, reliable and ideologically unbiased information to be able to choose the language they want to use with their deaf child and play a central role in his/her education: offering constant exposure to good language models in everyday face to face situations. Need to receive a complete representation of the language corresponding to their age; neither simplified nor limited in structure and vocabulary.

**\*In France, parents' language choice for their child in the context of education is a legal right (1). The method of language acquisition endorsed by ALPC (2) is French Cued Speech (LfPC). This is a method of spoken language acquisition using the "visual pathway" or "through vision"**

**Ideally:**  
Support & respect the parents' choice of educational project. With a parental educational project including Cued Speech adapted to their home language (LfPC for French language). The use of LfPC fulfills these needs, relying on the unimpaired visual sense, whatever the severity of the hearing loss, A very early exposure, before cochlear implantation helps develop a better reception of oral language.

**5. Follow Up**  
 Multi-disciplinary follow up: Speech therapists, hearing aid dispensers, psychologists, ENT doctors & geneticists, ENT reference center  
 Parents, still as natural language models and nurturers.

**Needs:** Need for a deaf person:

- to be literate and reach his/her academic potential.
- to be socially integrated with information totally made accessible in an environment where background noise makes hearing aids or cochlear implant inefficient (education, work, meetings).
- LfPC transliterators: to provide accessibility during the deaf person's academic career.
- to be in charge of their health path through child-adult transition project prepared by ENT reference centers

**Ideally:**  
LfPC helps deaf children to develop:

- a phonological awareness of the French phonological system based on syllables
- a precise representation of syntactic structures and connectors of the language
- the strategies involved in the reading process to become totally literate.

LfPC transliterators provide accessibility



\*Completion was led by Michel Francois (ERN CRANIO patient representative) from France who has two adult children with genetic hearing loss. Michel represents the patient organisation ALPC in France (Association nationale pour la Langue française Parlée Complétée). The content of this document has been validated by the board of ALPC. Content also considers information from French legislation and the French National Authority for Health.