



SURVEY ADOLESCENTS and ADULTS ANGELMAN

15 Years and more - April 2013 - France

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Introduction

Since the first description of the Angelman Syndrome in 1965 by Dr. Harry Angelman, Angelman adult world has been the subject of very few studies and we have hardly any information. By exchanging regularly with several families of adults, but also with young Angelman who question the future and development of their children, it seemed interesting to conduct a survey of families having an Angelman of 15 years and more.

From the youngest age as well as lifelong medical and educational monitoring allows one to mitigate certain manifestations of the Angelman Syndrome and to improve the lives of the Angelman. Awareness in the world adults, is to take preventive measures, because this is the youngest age that prepares adults for their Angelman life, but it also help professionals in institutions to better understand our children and therefore give them better support.

Our study sample : End of 2012, a questionnaire was sent by e-mail, or by mail, to Angelman families in France and abroad having a child 15 years Angelman and more, in order to develop a profile of the older Angelman with their features, their potential and their difficulties.

101 questionnaires were sent and in return **66 exploitable questionnaires** came back for:

- 24 women and 42 men
- Average age: 25 years (aged 15 to 55)
- 21 Angelman Syndrome aged 15-19 and 45 Angelman Syndrome 20 years and +
- Average age diagnosis: 9.5 years (less than 1 year to 40 years). For the 15-19 year olds, the average falls to 3 years, while for those without deletion of 15-19, the average is 6 years, and for 20 years and +, the average is 18 years.
- 43 with deletion, 13 with disomy, 7 UBE3A, 2 with lack of impression and 1 clinical diagnosis.

The number of people with deletion enters the range of % of deletion in the syndrome (between 60% and 70%) and, for ease of analysis, we regroup the people with other origins of the SA (disomy, UBE3A, lack of impression and clinical).

The results were divided by age group, by origin of the Angelman Syndrome (deletion and other genetic types (other)), and grouped in a general synthesis (total).

In order to make the reading of the document easier, we have grouped the results into a general summary, then the results were listed and detailed in 8 major topics:

1. Results survey - general information
2. Medical and paramedical
3. Traction and skeleton
4. Autonomy
5. Understanding and communication
6. Behavioural disorders
7. Activities
8. Facilities and support

1 – Result investigation - General:

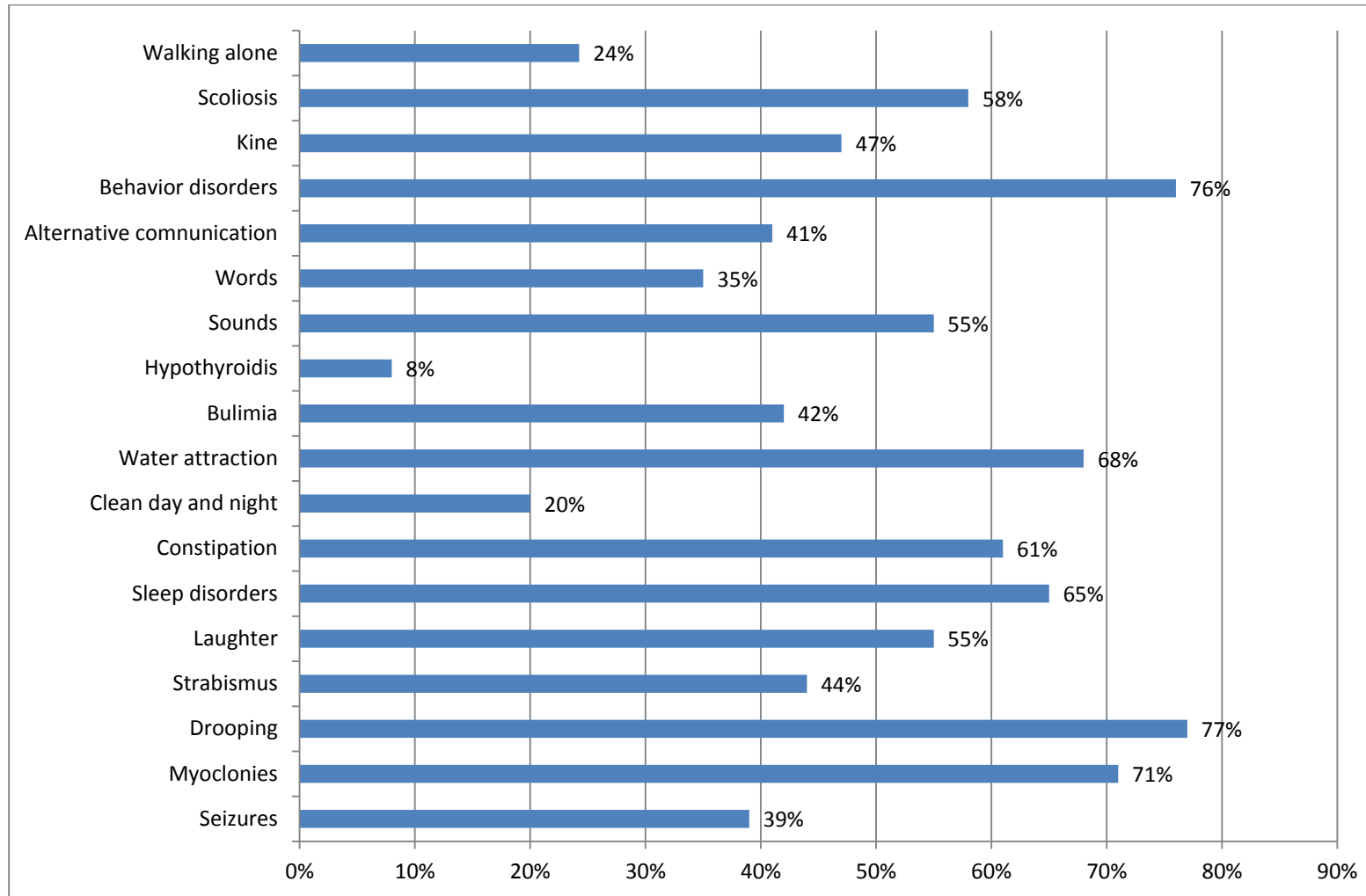
- **At the physical level: 24% walk unaided** (6 people have never walked), **58% have scoliosis** (especially deletions), but only 47% have a kine (in the more than 20 years, 25% go to liberals accompanied by parents). People without deletion are large in size and have a stronger body than those with deletion. There were two very tall people in the disomies.
- **Myoclonus** 71% of people (but only 10 per cent have a specific treatment).
- 39% have some active **seizures** only 5 people have never had such and 20% started to have them from 18 years). More than **90% are under antiepileptic treatment**. All genetic types are affected by crises.
- 42% are **bulimic** (61% of people without deletion) and it is difficult to manage this in the majority of cases. There is a resurgence of bulimia during the return to the parents homes of adults in boarding school, especially those with night bulimia cases.
- **Sleep disorders** are still 65% of Angelman despite an improvement in adolescence. There is often a resumption of unrest during an adult structured boarding school. Note is made that it is difficult to know how they sleep in boarding school.
- The problem of **constipation** is 61 per cent of all people having a diet or associated treatment. Some antiepileptic drugs aggravate this phenomenon.
- **Gastric reflux** is 21% of those with an associated treatment.
- **Laughter** diminishes especially among the more than 20 years without deletion; in some, laughing became rare.
- **The attraction for water** is always important, but decreases with age.
- **Hyperactivity** has disappeared (except for 2 people), and it becomes necessary to stimulate less eagerly in order to withdraw from Angelman.
- **Drooling** is present at 77%, but it had no treatment and rehabilitation.

- **Strabismus** is present at 44%, but it was only an operation, and **a single case of glaucoma** is observed.

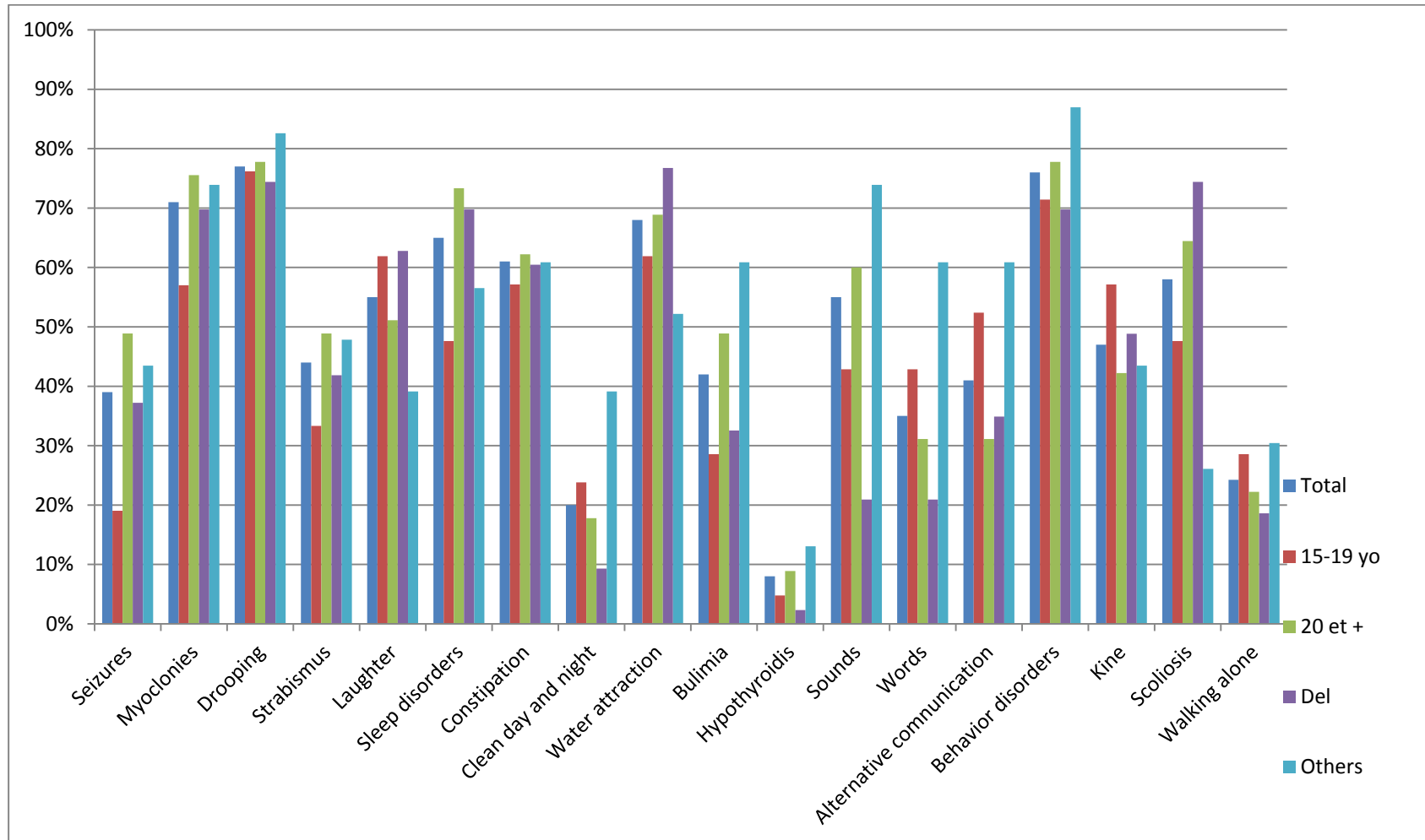
On the other hand, two remarks:

- on the way in which the Angelman looks at a document: they seem to need to scan the page
 - the fact they see wrong reliefs, which makes their approach more uncertain.
- Only 20% are **clean** day and night, and 33% during the day. Certain medications (antiepileptics or sleeping pills), as well as periods of crisis, worsen night enuresis.
 - **At the level of language and communication:**
 - 55% communicate by sounds,
 - 35% say words (especially in the disomies), but only one has a vocabulary of hundreds of words which are identifiable and appropriately used
 - 41% use alternative communication methods, especially those without deletion, and younger. Most are understood by their surroundings and have an understanding far superior to their expression (in any form whatsoever).
 - Progression remains constant, even beyond 30 years, and acceleration is often mentioned in the 18-20 years).
 - **Hypothyroidism** is only 8%, and remains at a fairly low rate.
 - **Behavior disorders** are important in 76% of cases and mostly affect those without deletion (87%). These disorders increased between 15 and 20 years, because the reasons for frustration are becoming more frequent and they have a more assertive character.

Figure 1 - General SA 15 years and more



Graph 2 - General by age and genetic origin.



2 - Medical and Paramedical

- **Constipation** concerns 61% and is accompanied by treatment in the long term and also a diet rich in fibre.
- **Epilepsy** is still at 39% of people despite treatment, with often a recovery around the age of 18-20 years, and also with the passage into adult structure. In 20% of cases, there is a worsening with age. **73% of those are under anti-epileptic treatment, 23% have attacks Myoclonic, 20% have absences**, and there are little generalized seizures. 5 people have never had a crisis.
- **Myoclonus affects 71 % of persons**, but only 30% have a treatment. Worsening is recorded in times of stress. It seems that this symptom is not properly taken into account by the medical profession, and it is very disabling for some people. The phenomenon increases with age and can have adverse effects on motor skills (walking and finer motor skills).
- **Bulimia** is 42% of people (61% of those without deletion), and requires the implementation of measures to put food out of reach (especially sugar) Behavioral methods have little effect, those involved always staying on the lookout. There is an bulimia during the return to the family home.
- In 45%, **delayed puberty**, with several cases of growth (11%) extending abnormally (up to 25 years) was noted. The girls all have their periods. 32% of males masturbate (usually in a reserved place). There was no search for sexuality with a partner, but friendships do exist.
- The **strabismus is present in 44%** of cases, and only 2 people have been operated
- Only three people over 20 years are followed by a **psychologist** and none among 15-19 year-olds.
- Few adults are regularly followed by **an orthopedic doctor** (30%).
- 9% have had **breathing problems**.

- **20% have reflux gastric** (with a resurgence at the age adult, and associated with behaviour disorders).
- **Scoliosis** requires a monitoring and control during the growth period. This topic will be developed in part motor.
- **Motor skills problems** worsen with age.

The difference of the structures of 15-19 years and 20 years and + results in a disparity of the supported in paramedical. This disparity is even greater if isolated "Cretons" (+ 20 years and place in adult structure pending): there are many more paramedical structures children - adolescents.

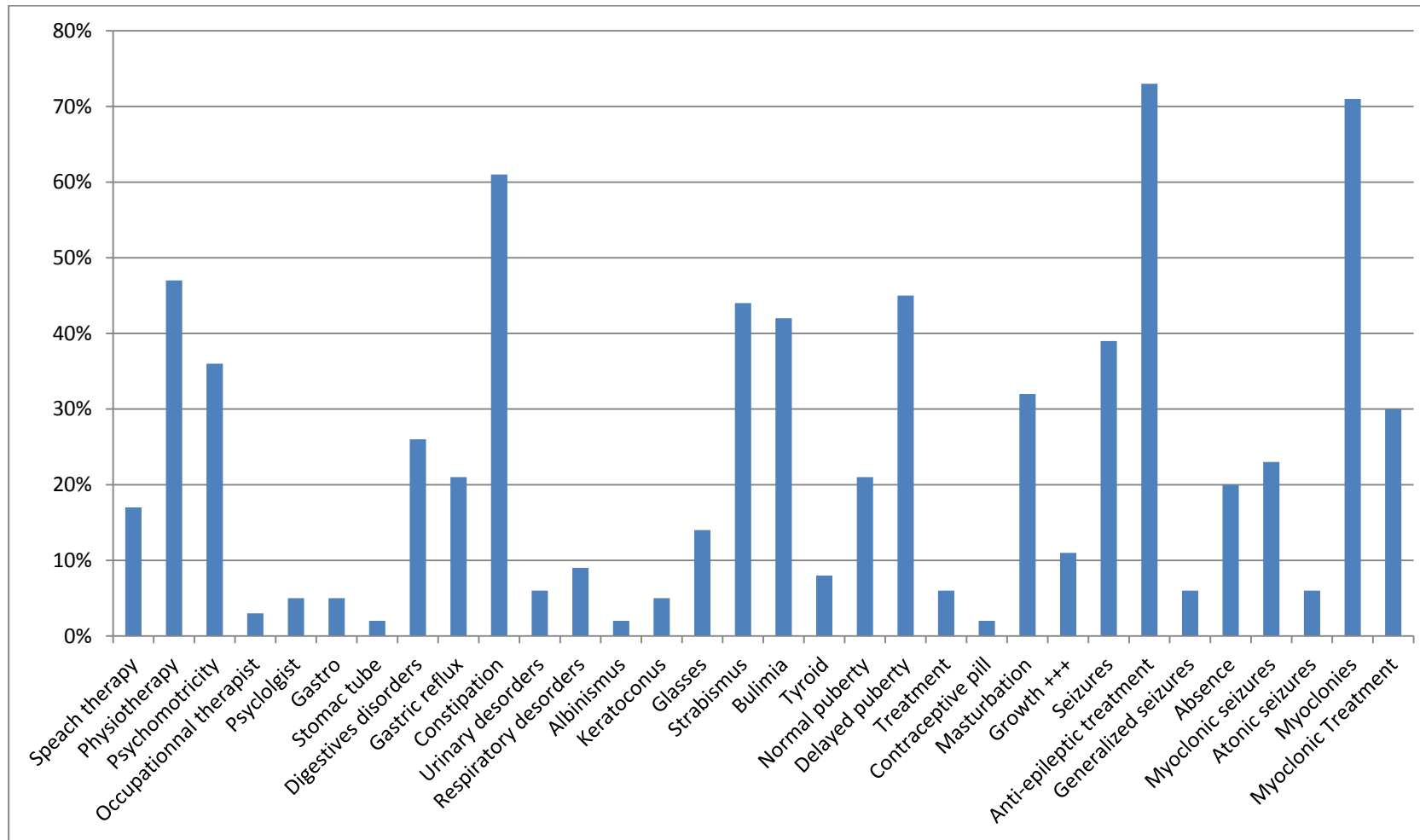
- **17% have speech therapy** (7% in the 20 years and more); very few have had the rehabilitation for the drooling (this is explained by the concept of non-verbal person so priority for re-education).
- **47% have a kine** (often a liberal with the family, aged over 25, for troubles with walking and folded position).
- **36% have the psychomotricity**, especially in the facilities for the polyhandicapped.

Comment: overall, it notes that the supported (even for children) needed much less nursing and with less equipment than those of young children currently.

Those supported require negotiations during the development of the individual project, and sometimes the parents fail to get them, thus they must opt for a follow-up in liberal outside the establishment.

The general State of health is good for 90%.

Graph 3 - Medical and Paramedical



3 - Motor and skeleton

Concerning motor global :

- **6% of people have never walked, 8% walk no longer and 30% have a significant regression.**
- **The average age of the acquisition of walking** in the Group deletion is 4.5 years (from 2 to 16 years); for others, the average age is 3 years (years 1-6).
- **73% walk only indoors and 44% walk whilst outdoors** . But only 32% are totally autonomous in their moving about.
- **14% are in wheelchair indoors** (5% for the 15-19 and 18% for 20 years and +) and **44% outdoors** (29% for the 15-19 and 51% for 20 years and +).
- **50% climb by themselves a staircase, and 25% can come downstairs without help.**

The problems at the level of the lower limbs (decrease in walking) are insufficiently detected and taken into account: only 47% have a kine (especially among 20 years and + always in child institutions type EMI and many families are forced to accompany their adult children to liberals because he has no support in institutions). It falls to 30% in more than 20 years. The follow-up to an orthopedic doctor is also low (between 30% and 50%). It would seem that professionals, and sometimes even parents, are not properly informed about the possible worsening during puberty (often up to 25 years).

In addition there is also a **greater tiredness** and a **decrease in physical activity**, which aggravate these phenomena. All categories of Angelman are concerned, including some people who walked very early and very correctly.

Only 32% have been fitted with **custommade orthopedic shoes**.

Angelman man 20 Years and +, took injections of botulinum toxin in calves (3 with success).

15% underwent operations at the level of the feet or knees, and 9% for scoliosis.

It was noted:

- **Retraction of the tendons** (32%),
- **A weakening of the knees** (52%),
- **Decline of the bust** (42%),
- **Deformations of the feet** (56%).

Scoliosis is found in 58 % people, mostly amongst girls and in the group with deletion.

It begins in 33% of cases before 12 years and 67% from 12 years. Going from 40% for those aged 15-19 to 80% in 20 years and + with deletion. **Which confirms that critical age is between 15 and 25 years.** It notes that, generally, the scoliosis stabilizes at the end of puberty. Only 14% have a corset (9% only at night), all in the deletions and 9% (severe scoliosis) had an operation with the correct results.

Important physical regression:


Total: 30% (including group deletion 35% and other 22%),

15-19 years: 14% in total (including group deletion 10% other 17%),

20 years and +: 38% in total (including group deletion 46% other 24%).

The problem of pain is not all mentioned in the complement of the questionnaires, while it can be the cause of a refusal to walking.

Summary mobility and skeleton (next page)

	TOTAL	Del	Other	15-19 yo	20 yo et+
To only hold sitted	89%	86%	96%	95%	87%
To hold sitted with assistance	11%	12%	9%	5%	13%
To crawl	47%	40%	61%	33%	53%
To go to 4 legs	44%	40%	52%	62%	36%
To only put itself upright	64%	47%	96%	67%	62%
To put itself upright with assistance	27%	40%	4%	29%	27%
To only go	73%	63%	91%	52%	82%
To go while being held inside	29%	35%	17%	33%	27%
To go while being held outside	44%	56%	22%	57%	38%
Never walk	6%	7%	4%	5%	7%
Do not walk	8%	12%	0%	10%	7%
Facilitator	6%	9%	0%	5%	7%
Armchair inside	14%	16%	9%	5%	18%
Armchair outside	44%	51%	30%	29%	51%
To climb alone	50%	35%	78%	48%	51%
To climb with assistance	38%	42%	30%	43%	36%
To descend the staircase alone	38%	26%	61%	38%	38%
To descend the staircase with assistance	39%	37%	43%	38%	40%
Retraction of the tendons	32%	37%	22%	33%	31%
Bending of the knees	52%	53%	48%	48%	53%
Bending of the bust	42%	47%	35%	48%	40%
Deformation of the feet	56%	60%	48%	57%	56%
Toes out of claw	15%	19%	9%	14%	16%
Ortheses	14%	12%	17%	24%	9%
Orthopedic shoes	32%	35%	26%	33%	31%
Orthopedic soles	20%	19%	22%	14%	22%
Botulinic toxin	6%	5%	9%	0%	9%
Operation of the feet or knees	15%	16%	13%	10%	18%
Scoliosis	55%	67%	30%	48%	58%
Surgery	9%	14%	0%	10%	9%
Girdle maintenance	3%	2%	4%	0%	4%
Corset day	14%	21%	0%	14%	13%
Corset harms	9%	14%	0%	10%	9%
Important physical	30%	35%	22%	14%	38%

Fine motor skills:

95% of the Angelman have important, coordination difficulties often related to fine motor skills problems and learning difficulties: for example difficulty to hold and properly use an object, too many distractions, difficulty to fix his attention.

- 32% are able to play ball (throwing and catching),.
- 48% are able to follow objects, 39% screw, 21% use scissors (one can cut his nails),.
- 55% are able to scribble or draw.

Motricity at meal table:

- **45% of people eat by themselves**, but however, there is a significant difference between the two groups: the Angelman with deletion have more difficulty to eat by themselves and 74% of them need assistance to eat (compared with 17% for others).
- However 63% arrive to eat with their hands: it is the use of a knife & fork that causes a problem.
- 71% are able to eat with a spoon.
- 71% are able to drink alone (mostly with a normal glass).
- 14% use a table knife (but only one person is capable of cutting meat).

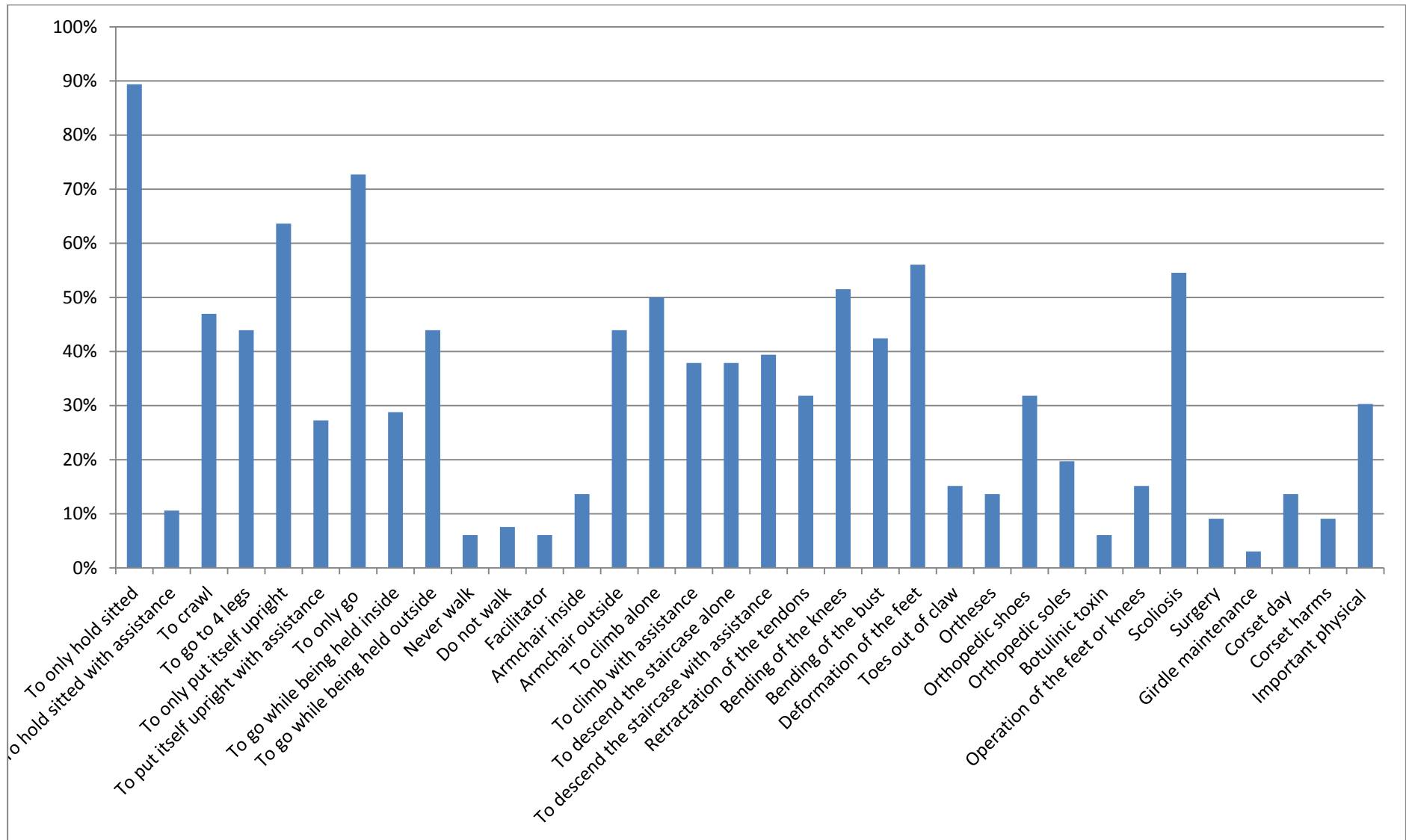
It also notes that the Group of 20 years and + has more skills. This is explained by the long learning and improvements that occur after 18 years. One can also wonder about different learning proposed over the years.

It is worth noting that some people can perform any of these very important activities due to myoclonus, while they were younger: inability to drink, bring food to their mouth,...

Summary table:

Fine motor skills					
	TOTAL	DELETION	OTHER	15-19 years	20 years and +
Difficulty of important coordination	94 %	95 %	91 %	95 %	93 %
Catch a ball	32 %	30 %	35 %	24 %	36 %
Throw a balloon	41 %	40 %	43 %	38 %	42 %
Put into boxes	48 %	35 %	74 %	52 %	47 %
Screw	39 %	30 %	57 %	43 %	38 %
Cut with scissors	21 %	9 %	43 %	19 %	22 %
Thread beads	33 %	21 %	57 %	38 %	31 %
Scribble	55 %	40 %	83 %	52 %	56 %
Eat by themselves	45 %	28 %	78 %	43 %	47 %
Eat with help	55 %	74 %	17 %	57 %	53 %
Eat with hands	45 %	63 %	13 %	57 %	40 %
Eat with a spoon	71 %	60 %	91 %	67 %	73 %
Eat with a fork	62 %	47 %	91 %	62 %	62 %
Eat with a knife	14 %	2 %	35 %	5 %	18 %
Drink by themself	71 %	60 %	91 %	76 %	69 %

Graph 4 – Mobility and skeleton



4 - Autonomy

36% of families consider that their Angelman over 15 years of age has a very reduced autonomy (of which 58% among those with deletion).

We have seen in the paragraph on motor control, certain disorders which brake the autonomy of the Angelman:

- **14% do not walk** (or more), **44% must be helped whilst outside** and **50% need help on stairs**.

We now discuss the autonomy in the tasks of everyday life: most are in need of help for all acts, some have no autonomy, others (rare) arrive to 'fend' alone, but always under supervision.

Many like to participate and manage to perform a few simple tasks, albeit imperfectly. And they are very proud to be useful.

- **53% eat by themselves** (must cut their food, with a single knife)
- **30% can serve themselves**,
- **62% can drink by themselves**
- **41% participate in regards to the meal** 41% lay the table, 33% clear the table and 24% wipe the table, 14% fill and empty the dishwasher, 6% arrive to cut and peel the vegetables,
- **32% undress by themselves** , and 29% with help,
- **42% dress with help** (one dresses by himself), difficulties due to the fine motor control (buttoning, lacin, tightening...), but also recognize the place of WS. However, some arrive, casual, to dress properly,

- 70% **go to the toilet**, but 39% **wear diapers by day** and 80% **at night**,
- 39% **wash by themselves** (under supervision),
- 18% **choose their clothes**.

It is worth noting that:

- 21% **must be stimulated**,
- 30% **manage their free time**.

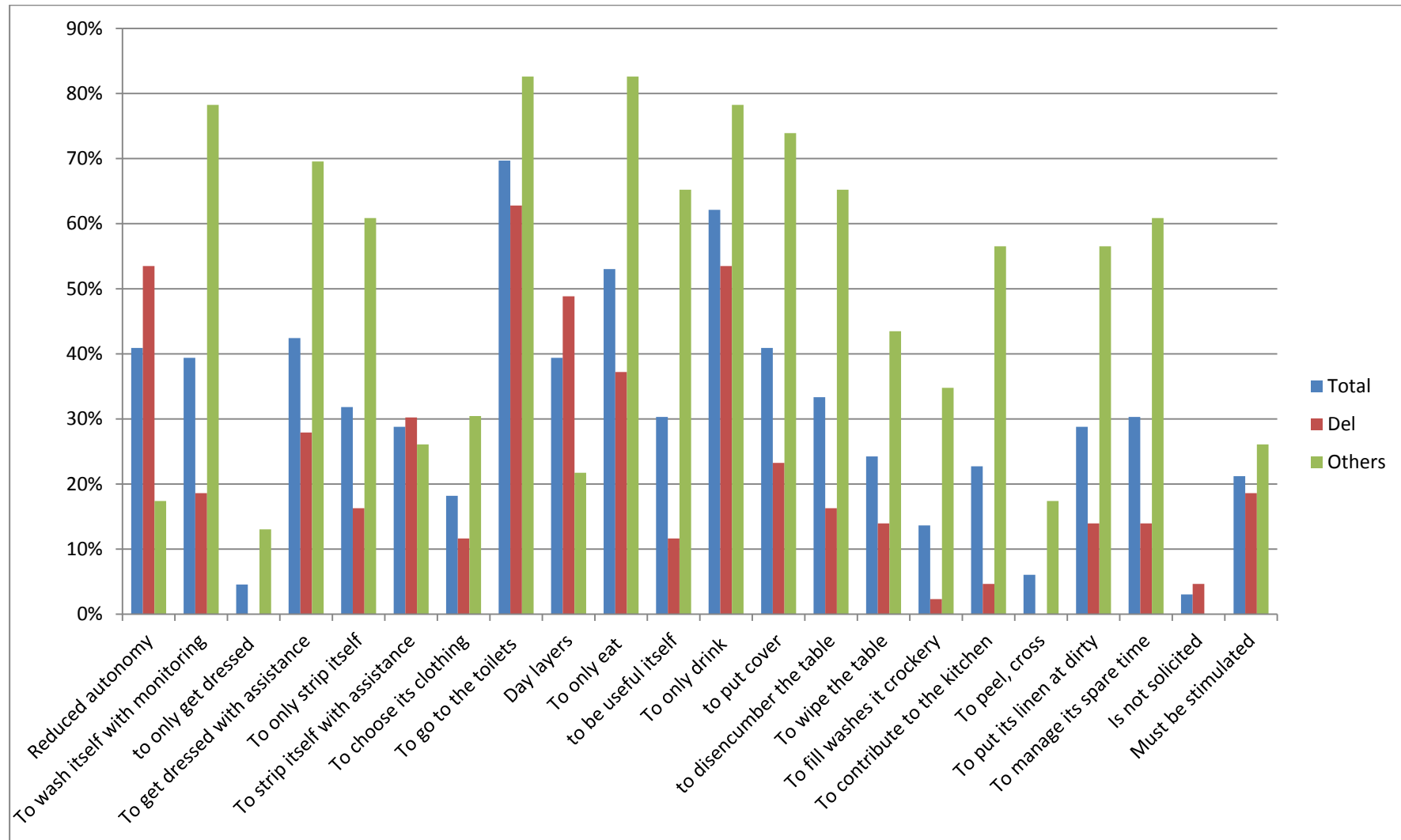
This lack of autonomy brought 47% of families to arrange their homes, and 20% are planning to do so soon (changes are made as the difficulties encountered):

- 9% **have completely adapted housing**,
- 11% have **handrails**,
- 18% **have access by ramps**,
- 42% **adapted bathrooms** (70% of the families assume by themselves the toilet),
- 14% **have a suitable bed** (essentially a nursing bed and no beds with barrier),.

And 9% **have a vehicle equipped** (ramp or lift).

Two adults have a guide dog.

Graph 5 - Autonomy + 15 yo



5- Understanding and Communication

Communication difficulties are an important element in the Angelman Syndrome. Most people are able to understand simple orders as part of their daily routine. But all have difficulty understanding complex orders and all orders which are abstract.

- **91% of the Angelman are eager to communicate** despite verbal impairment, some manage to impress people who are strangers to them, **74% provoke the encounter with the other**, which is facilitated by their jovial appearance. This social interaction is often excessive and disorderly, as they can disturb others by seeking at all costs attention and grabbing people. If older, the fear of the stranger seems to disappear, some have a fear of crowd and noise, which disrupts their communication and often translates into shouting or uncontrolled laughter.
- Oral language level: 27% are able to say a few words (including three about ten and one adult is able to use more than 300 words), 52% use sounds and 14% use meaningful syllables and sounds. 13% of adults associate words.
- **53% are able to communicate by a language that is their own** (sounds, simple gestures, photo album, postures, pointing of the finger (35%), take you by the hand (11%), but also combining gesture and Word or sound...).
- **38% have had access to the alternative communication** 43% among the 15-19 years. 29% use some simple pictograms (one uses 30).
- **62% are able to make a request** : drinking (47%), eating (45%), going to the toilet (35%), an object (39%), activity (29%), see someone (23%), and therefore to make known their needs.
- **77% understand simple instructions, 32% follow a conversation, 62% understand words.**
- **37% respect the rules of life** (even if callbacks are required).
- No-one accepts not being understood and all, when this happens, manifest their dissatisfaction.
- Only 11% manage their security, and in a suitable environment.

This understanding is improving with age, because they are calmer and better arrive to fix their attention:

They are able to focus on a longer period: 40% have more than 15 minutes attention, some manage by themselves for more than an hour and only 18% are unable to focus their attention. 27% of adults are able to wait if one warns them.

We can consider that **laughter is an element of communication** which must not be overlooked. It is related to the context, and not necessarily in joyful situations; Sometimes it shows anxiety, fear, excessive excitation.

- 90% are curious and looking for noise.
- **72% are able to make choices**, for food, clothing, games,...
- **9% recognize words, 23% graphics,**
- **22% recognize colors,**
- **54% scribble,**
- 54 % are able to undertake a simple activity**
- **45% take initiatives,**
- **35% have a repetitive questioning** despite the replies,
- 85% know how to move indoors and 43% outdoors.

The proposed support must therefore take into account all these elements: pictured here is the importance of the follow-up by a speech therapist or educator specialized in communication, and the essential link between professionals and parents to go in the same direction:

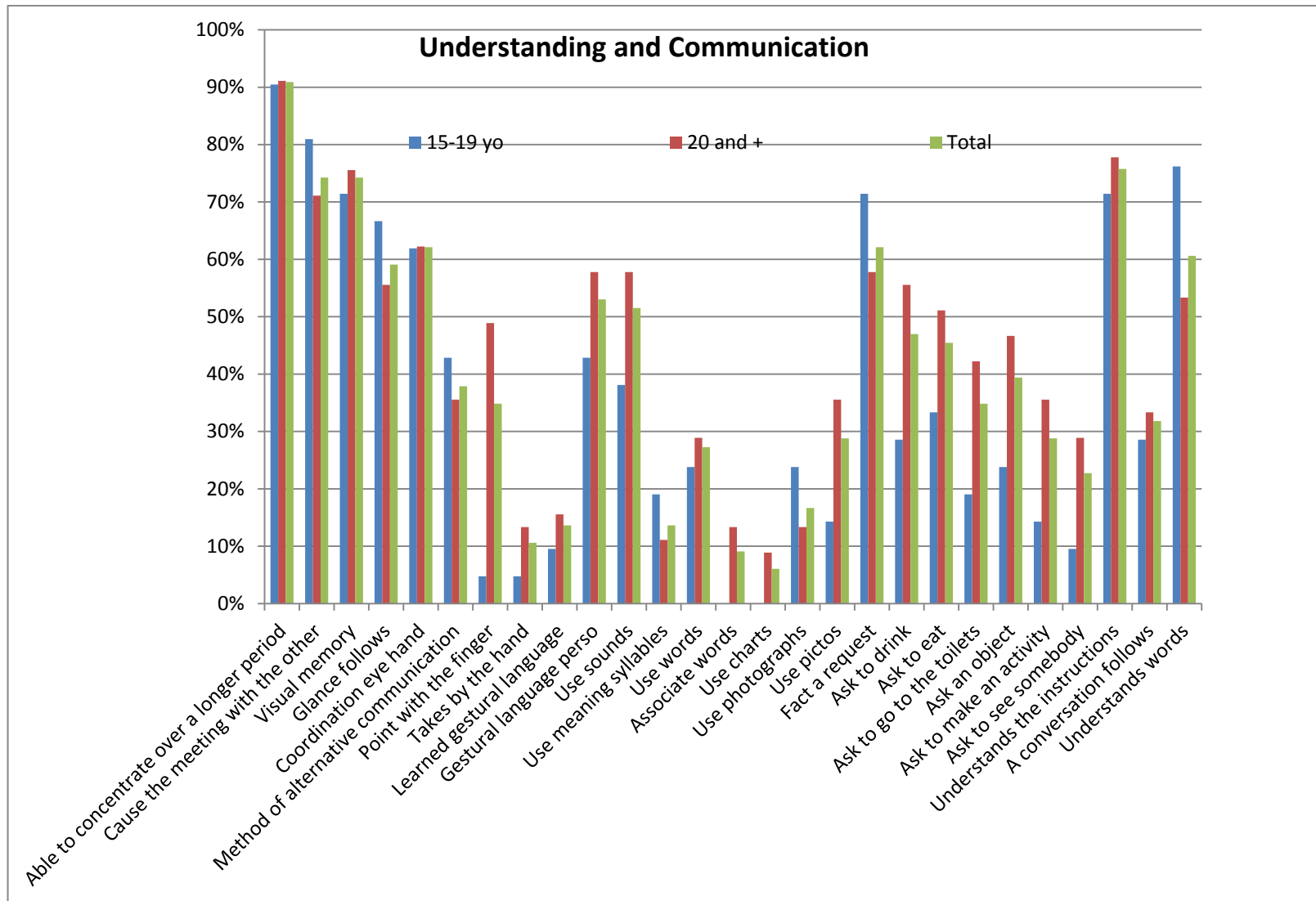
- Importance to **help them find their own way of communication and the suitable supports,**
- **Help them keep what they have found,**
- especially **the communication tool between the establishments for adults and families,**

Some began understand successfully at adulthood alternative communication tools and continue to progress.

Understanding and communication

	15-19	20 et +	Total		15-19	20 et +	Total
Able to concentrate over a longer period	90%	91%	91%	Ask to eat	33%	51%	45%
Cause the meeting with the other	81%	71%	74%	Ask to go to the toilets	19%	42%	35%
Visual memory	71%	76%	74%	Ask an object	24%	47%	39%
Glance follows	67%	56%	59%	Ask to make an activity	14%	36%	29%
Coordination eye hand	62%	62%	62%	Ask to see somebody	10%	29%	23%
Method of alternative communication	43%	36%	38%	Understands the instructions	71%	78%	76%
Point with the finger	5%	49%	35%	A conversation follows	29%	33%	32%
Takes by the hand	5%	13%	11%	Understands words	76%	53%	61%
Learned gestural language	10%	16%	14%	Can wait	19%	27%	24%
Gestural language perso	43%	58%	53%	Do not fix its attention	24%	16%	18%
Use sounds	38%	58%	52%	Attention duration - 15mn	38%	47%	44%
Use meaning syllables	19%	11%	14%	Attention duration + 15 mn	48%	36%	39%
Use words	24%	29%	27%	Recognizes written words	5%	11%	9%
Associate words	0%	13%	9%	Recognizes graphics	19%	24%	23%
Use charts	0%	9%	6%	Recognizes the colors	29%	18%	21%
Use photographs	24%	13%	17%	Gribouille	48%	56%	53%
Use pictos	14%	36%	29%	To make choices	81%	67%	71%
Fact a request	71%	58%	62%	Repetitive questioning	29%	38%	35%
Ask to drink	29%	56%	47%				

Graph 6 – Understanding and communication



6 - Behavior disorders (see figure on next page)

The first element of observation is the **net decrease in hyperactivity** that passes from 27% in the child to 6% in the adult Angelman. The 20 years and + are less prone to excitement than the 15-19 years. Laughter is less present and sadness may appear on their faces. On the other hand, they become slower and less active, and it becomes necessary to significantly motivate them to bring them out of their shells (they love staying lying down, watching TV). More than 10% refused to walk and go outside. Adults are obstinate, and it is difficult to make them change their minds.

62% of the families interviewed are part of frequent behavioral disorders (largest in the group without deletion), and only 11% believe that their child has no disorder.

The causes of these disorders are usually identified (in descending order of frequency):

- Expressive communication problem,
- Refusal of an application (often related to food),
- Problem of understanding,
- Interruption of a valued activity
- Lack of anticipation and confusion in the situation, waiting,.
- Unpleasant sensory simulation,
- Pain.

The manifestations of these disorders are often specific to the Angelman Syndrome (by decreasing frequency):

- Excessive laughter (often a laugh, nervous and unstoppable, described as scary by some parents),
- Shouting (often very loud and long-lasting),.
- Throwing of objects (which are handy),.
- Anger,
- Hair-pulling, pinching,
- Violence on themselves or their clothing,.
- Opposition, throwing themselves on the ground,
- 26% **self-absorption** with a situation difficult to manage.

Trends become present from the earliest age in some cases, **bulimia** becomes worrying in 61% of adults without deletion (especially UBE3A and Disomy) who will be in constant quest for food and have overweight problems. They usually have a diet, but require constant monitoring. Some get up at night to eat when they are back in the family home.

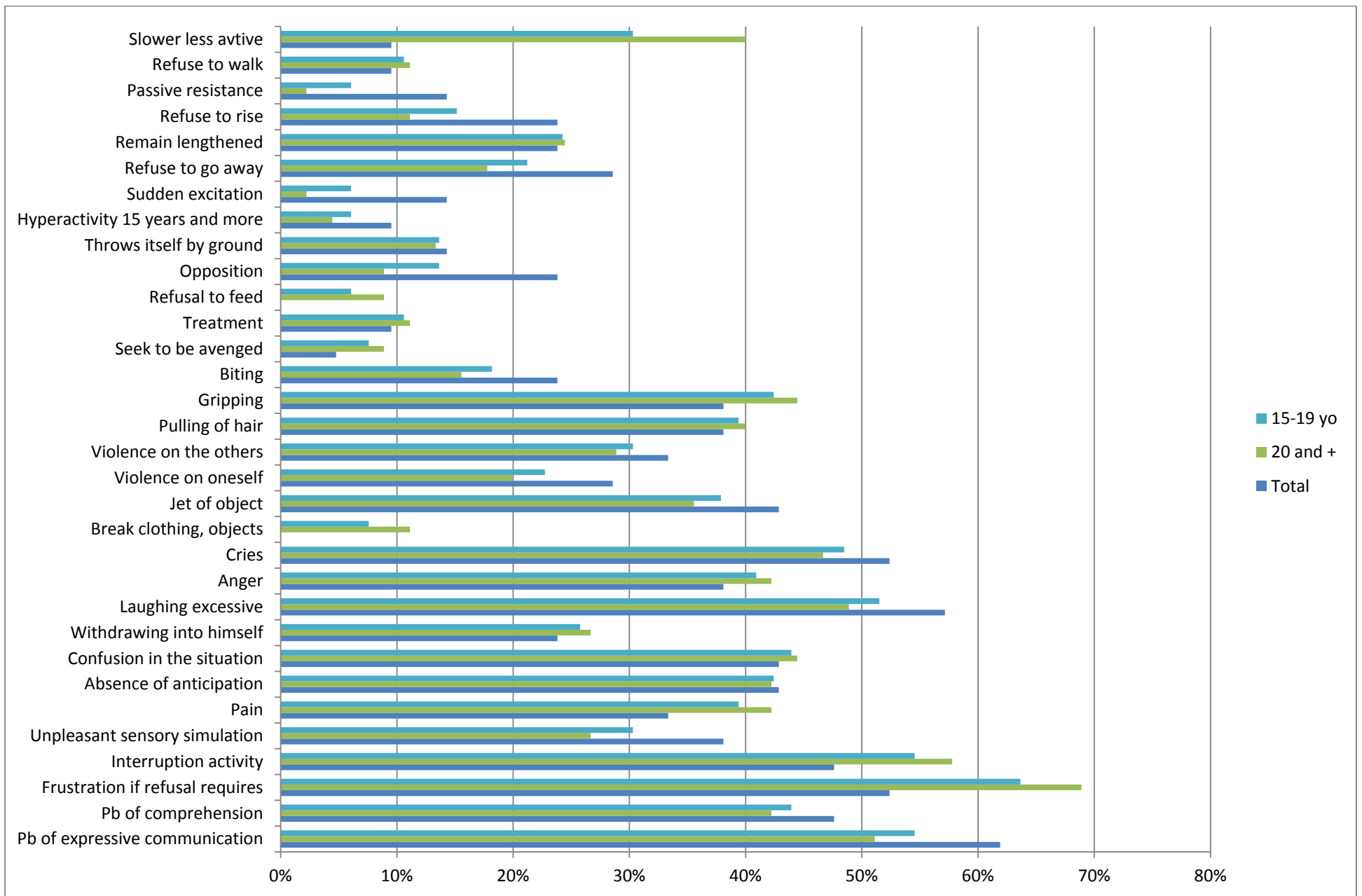
Conversely, some refuse to eat when going to a boarding school and / or adult structure.

Support for these disorders:

- Not really appropriate care, little is proposed for behavioral therapy. Note that only 4 families have received training on the management of disorders of behavior, given by a Neuropsychologist. This allowed them to have a better understanding of the functions of their children, of reasoning compared with maladaptive behaviors, to play down certain tensions. Three institutions manage these behaviour problems with an improvement seen by parents,
- One adult has a weekly follow-up with a psychologist at the request of the parents,
- Two with a psychiatrist,
- **11% have one or more treatments** (reported drugs: Tercian, Atarax, Notizan, Neuleptil, Risperdal, Gardenal, Effortil, Lysanxa, Loramet, Doctrazone and Tegretol).

When entering into the adult structure, these behavioral disorders tend to grow for the first months, with sometimes an overmedicalization that families are forced to manage with the help of neurologists or family doctor (some medication reduces daytime alertness and resulted in difficulties at the level of mobility).

Chart 7 - Conduct disorder (see next page)



7 - Activities

These **activities are sometimes short-lived**, lack of patience or attention (44% less than 15 minutes and 39% between 15 minutes and an hour), and 18% are not capable of fixing their attention at all. Physical activity decreases with age due to greater fatigue and the desire of some to remain quietly lying down.

- 24% are able to wait calmly, but impatience is the rule.
- 18% of adults are able to time orientate themselves (after now, today ' today-tomorrow, short-long). Some connect time with photos, another has a calendar and marks it every day to get a picture of what will be happening the next day. The use of a timer allows to time activity or to wait before an activity.
- 39% are able to undertake a simple activity and deal with it themselves.
- 44% are capable of initiative and ask to do an activity.
- 21% can stay alone in a room if they are occupied (an adult can stay alone for half an hour in being held accountable). The majority need constant monitoring and are on the lookout for an "entrepreneurial freedom" not always to the liking of the parents. They enjoy with extreme quickness when vigilance is reduced to indulge in their favorite activities with their own rules (parents busy on the phone, talking, cooking,...). The ban is known, but not met in these moments.
- 11% are able to maintain their security, and only 2 adults are able to manage their free time.
- 83% are able to move indoors, and 42% outside.
- 55% require repeated calls to groom themselves, because the attraction to water has decreased.
- In 30% of cases, it is difficult to stop an activity, and parents should anticipate.

List of activities carried out with the family:

- Listening to music (CD, Youtube, etc): 64% watch the TV or video 85%,
- Educational games (puzzle, memory, fishing line,...): 47% aged 20 and +,.
- Painting, drawing, doodling,
- Making music,
- Computer or iPad (internet, Youtube, gaming applications, educational applications),
- Reading cooking magazines, catalogues of supermarkets, books (but also for some to cut or rip them very carefully,
- Playdough,
- Gardening,
- Walking with or without a wheelchair, often with a specific purpose; many adults are still able to do several kilometres by walking,
- Drive by car, by boat, by train, by public transport,
- Tricycle, tandem tricycle in duo, (an adult has normal bikes), swing,
- Ball games(by foot or hand), balls, rackets, petanque, ping-pong, (sometimes activities are made sitting)
- Play in or with water (but decrease with age): pool, beach, «Tiralo», wading in the water,
- Horse riding and horse-drawn carriage,
- Eat and go to the restaurant, to the Mac do, an aperitif with friends...

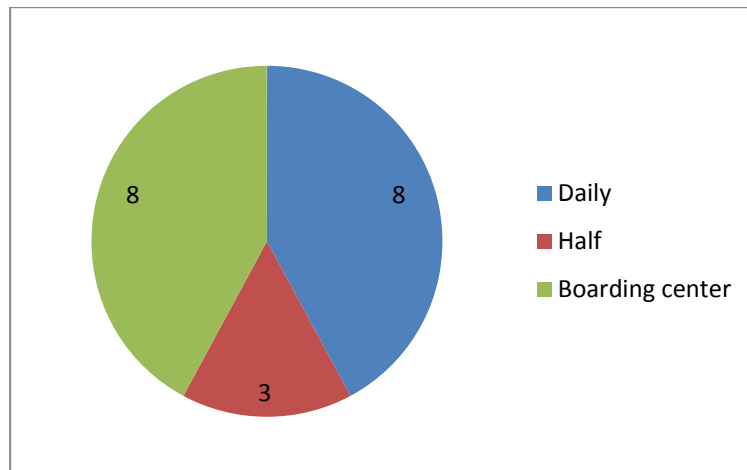
8 - Host institutions and support

8 1 - The 15 - 19 Years :

The Angelman Syndrome this age group 21, 19 are in structure children-teens type IME, and 2 at the home of the parents.

8 are close to their home, 3 have more than one hour of airlift.

Figure 8: operating home of the 15-19 age (number of persons):



- This home is in **74% to waiting parents**. But most noted a decrease in activity with increasing age.
- **26 %** relatives believe that the **activities are inadequate**, and 11% have trouble to obtain information.

The proposed activities:

- 84% balneotherapy (not always on a regular basis),
- 68% music,
- 63% frequent trips outside,
- 58% drawing, painting,
- 47% horseback riding or horse-drawn carriage
- 26% video, cinema,
- 25% tricycle,
- 16% gardening,
- 11% relaxation, 5% computer or iPad
- 5% games,
- 5% tales, singing,
- 5% sport,
- 5% kitchen.

Paramedical activities:

63% physiotherapy, 37% speech therapy, and 47% psychomotricity

There is a lack of physical activity, and some parents are obliged to conduct themselves their children to the pool and the physiotherapist. There is no awareness of the risks of scoliosis and worsening of the Motricity at puberty. The Angelman Syndrome could benefit from the stimulation of physical activities in the group.

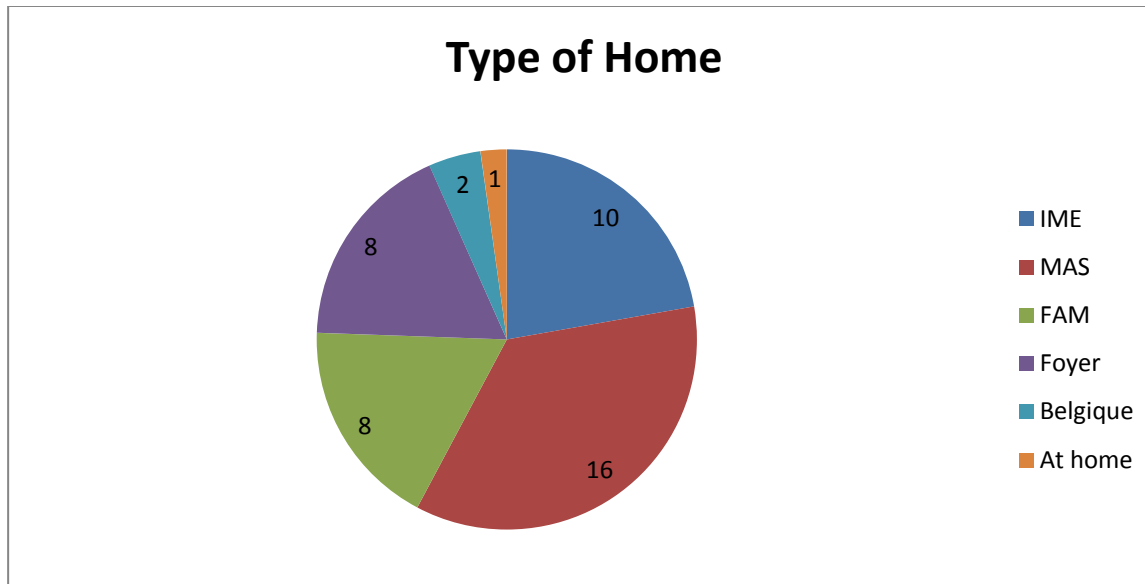
Also, speech as well as the alternative communication methods are not the general rule. The Angelman are not priority insofar as they are considered non-verbal. Only institutions that primarily receive autism offer residents a communication aid.

8 2 - The Angelman Syndrome 20 years and +:

The passage into an adult establishment is planned for the age of 20 years, but the lack of place delays this passage:

23% of adults are still institutionalized children (Amendment Creton, which requires institutions to keep children as long as there is not a suitable place). Some IMEs create "pre-adult" groups which gather young people from 20 to 25 years. Indeed, the facilities and the support are not really adapted, and cohabitation with younger children is not always obvious.

Figure 9: Type of home for 20 years and more (by number of persons (total 45)): We can state that out of the 32 Angelman placed in adult institutions in France: 50% are in MAS.



Out of the 45 Angelman of 20 years + :

- 10 are still institutionalized children (type IME) i.e. 23%,
- 16 are in Specialised and very medicalised Homes (MAS) or 36%,
- 8 are in Medicalised Foster Homes (FAM) or 18 %,
- 8 are in Home Center or 18 %,
- 2 are in an establishment in Belgium,
- 1 is at the home of the parents waiting for a place in the MAS.

Of the 44 Angelman aged 20 and +, and institutional :

- **27 Angelman with deletion** : 7 are IME, 13 in **MAS (48%)**, 13 4 FAM, 3 in a home life.
- **17 Angelman with another genetic type** : 3 are IME, 3 in MAS, **4 in FAM (24%)**, **4 in home life (24%)** and 2 in Belgium.

The distribution is therefore significantly different.

- **75% are boarding** and 25% in semi internship.
- Going outside : **42% every week** , 36% each fortnight, 9% each month, further away, 6% 3% on-site visit.
- The boarding school was half imposed or chosen by the families.
- 50% are close to home, distant 30% and 20% very distant.
- Average age of residents in groups where adults are welcomed: 20% under 30 years old, 50% between ages 30 and 45 and 30% more than 45 years.
- The adaptation has been easy in 50% of cases (less than 6 months), average over 24% and difficult in 26%.
- 50% of families say the establishment corresponds to their expectations and 34% not at all. Most often, the parents find a lack of mental activities as well as physical.

List of the activities proposed, all types of establishments confused (but often without regularity):

- 64% balneotherapy,
- 64% music,
- 57% drawing, painting,
- 55% video, cinema,
- 50% riding or horse-drawn carriage
- 48% frequent outputs,
- 30% gardening,
- 23% computer or iPad
- 23% sport (cycling, tricycle, ball),
- 23% games,
- 16% kitchen,
- 16% relaxation,
- 11% aesthetic,
- 11% tales, singing,
- 9% domestic activities,
- 9% pet therapy.

27% were of **physiotherapy**, including 11% in liberal (only 18% among those without deletion); 7% have **speech therapy** (only 2 Angelman with deletion), 41% of the deletions are followed by a **psychomotrician** versus 18% for others), one is followed by a **psychologist** and one by an **occupational therapist**. Results are really very low, and give concern for the future.

Many parents try in vain to get a better support in institutions for adults, but often see opposing concepts of severe disability and non-verbal.

There is **a huge lack of information within institutions for adults Angelman syndrome, a lack of awareness of the possibility of evolution to adulthood of the Angelman and also the great difference between their understanding and their verbal expression.** Some adults are regarded as aggressive while: either they are simply not understood and seek to communicate, or lack of activity translates and occurs a hyperexcitement .

It is in MAS that there is the least amount of proposed activities, there is much going on in care and nursing and not enough in the activity. This is due to the Organization and a low emphasis on activities.

60% of the Angelman are found mixed with more disabled people and it is therefore difficult to find appropriate activities for all. It is usually in collective activities without looking for progress and a lack of educational activities. Individualized activities remain exceptional and are of short duration. Only 5% of the Angelman are associated with domestic activities.

- 15% of families totally lack information on the activities of their child.
- 50% have difficulty knowing what is actually done.
- 40% of the Angelman have very irregular activities, because there is no existing or respected programme.
- In 70% of institutions, there's no separation between the areas of daytime and nighttime. Very few have workshops for activities in reserved rooms.
- The activities take place most often from 11: 00 to noon and from 3 pm to 4 pm. The rest of the time is dedicated to body care and resting. *It is rare to find planned evening activities.*

Out of the 16 Angelman cared for in MAS:

Only 3 people have regular and varied activities. But it is worth noting that some Angelman remained purely as spectators and thrive very well of this lack of activity. Some have many more other activities within the family.

- 95% have aesthetics and relaxation,
- 4 go out more than twice per month,
- 6 have a painting activity,
- 6 have balneotherapy,
- 5 have a musical activity,
- 4. go horse riding,
- 4. do gardening,
- 15 have television periods

On the 8 Angelman cared for in FAM:

3 have very varied & rewarding activities with a fixed timetable which is respected. The use of alternative communication is promoted, the individualized project concrete and respected. Support is scalable and adapted to each.

For others, there are more activities and are more varied in MAS activities. Because these institutions are more recent, the population is less aged and the staff are younger. Finding more leisure activities, and more socialization.

On the 7 welcomed home life:

4 have activities adapted with respected individualized projects, but few physical activities. Given the aging of the residents, some homes are considering a conversion into FAM to integrate a better follow-up medically and paramedically both falling under the liberal home.

The 2 adults taken in charge in Belgium combine all activities. Parents are totally satisfied with the support, even if they regret the remoteness that reduces the time with the family.

Adult in-home family (by choice) is waiting for a placement in MAS, because it becomes difficult to manage a constant presence.

Conclusion and summary:

We have seen, throughout the investigation, given all the genetic mechanisms to the Angelman Syndrome a clinical picture of significant intellectual impairment, with movement disorders, characteristic behaviors and an important limitation of communication and language. But that, as demonstrated by recent studies, we observed that some clinical differences are correlated with the genotype. And within these different genotypes exist significant gaps.

People with disomy fewer larger stature, less poorly controlled movements, ataxia. As well as those who have a lack of markers, they have a more developed language ability. As the UBE3A, they tend to be the most important bulimia. The group without deletion is easier with regard to fine motor skills.

But 95% of Angelman within our study have the same behavioral characteristics, and disorders of the market (with contractures of the members), sleep and epilepsy continue to be important in the life of the adult Angelman.

This observation shows a great disparity in the population of the Angelman Syndrome which must absolutely be taken into account in medical followup, the therapeutic and educational, supported by the guidelines in specialized institutions.

A few trends emerged from our study :

About Motricity:

- There is much worsening of some general motor skills from the age of 16-17 years, and in others with the appearance of scoliosis; with a stabilisation at the end of growth. Scoliosis, or curvature of the spine may require an operation that will improve the quality of life.
- Early and intensive learning are not associated with better motor skills in adulthood. Some have walked very early and have big problems in adulthood. On the other hand, the market should be encouraged. Weekly physiotherapy sessions must be proposed to combat the decline in muscle tone (more stiffness and bent members). The size and weight of adults have often been mentioned as factors that complicate support overall, both at home and in institutions. We must think hard to encourage this through for example : traction to swimming and dancing, more playful than the market.

- The use of the wheelchair outdoors does not reduce the capacity to the market of the Angelman, but longer trips, both long and secure (note that it is frequently used to better manage outings into shops, exhibitions, fairs, integral part of the socialization and are often very appreciated).
- The wheelchair outdoors (51% in the group with deletion) is mainly used for some long trips, either due to the fatigue or because of refusal to walk long distances. The majority of the Angelman is in trouble on rough terrain, and one has the impression that they do not properly see the obstacles, which causes frequent falls.
- Also note that due to seizures with a brutal fall, some are constantly under close surveillance (tripping over an obstacle may be sufficient to trigger a crisis) and that some persons may not perform any of these very important activities due to myoclonus, while they were younger: inability to drink, bring food to their mouths.

Concerning the cognitive and communication:

- A positive development in the cognitive field and which has also been observed in the communication. The Group of 20 years and + have more skills. This can be explained by their better concentration that promotes learning often at the end of the growth period. Hyperactivity, pushing them to be always agitated, decreased and this decrease did not always diminish in them the curiosity and the desire to have activities, while helping them to ask more. In addition, the more older were often enrolled in kindergarten and much later joined the world of disability.
- The understanding, communication and learning continues to evolve, with an acceleration from 18-20 years. It must be taken into account this development when structuring for adults. In the majority of cases, people have their own type of communication that combines words, sounds, gestures, pictures and pictograms. Motor difficulties are learnt gestures which are often distorted and difficult to understand. Many find it easier to use their own actions more in relation to their motor skills. The use of pictograms is reduced to 10 maximum, probably because of the difficulty for the Angelman's access to the abstract. On the other hand the use of pictures to make a request, a choice, seems easy with a sometimes slow start. At any age, it is possible to successfully introduce new methods of communication and the use of new technologies.

- It is important not to reduce the communication to food and animals, because if **the Angelman have difficulty to speak, they have a lot of things to say**. And please take the time to listen and let them express themselves. It is also important to transmit to all carers or professionals, the sounds, syllables and the gestures used thanks to photos (for actions), a list written for what is verbal, the symbols and their meaning for the person. And always ensure by rephrasing that it has been understood.

This positive development has nevertheless its corollary :

- The desire for autonomy that growing up that produces, and this is normal, the transition to adulthood, and the affirmation of the personality and the character. It becomes more difficult to manage the behavioural disorders because of the size and corpulence Angelman, disorders that are amplified weigh heavily with regard to parents, aging themselves, and accompanying them.
- Actions that make a child smile become difficult to manage adolescents or adults; It therefore seems important to act against these disorders from the earliest age. The Angelman are stubborn and like to do things in their own way...
- The proposed activities must be adapted to their age and their capacity. Some were able to continue their leisure time with their favorites with adapted to overcome poorer motor skills (for example, certain physical activities can be done while sitting (think to remove the armrests), or just sitting down on a stool at their height: ball, balls, ping-pong, basketball hoop, the tricycle in duo, a Chair swing rolling, there are floating bathing suits), you can fit an engine under a manual wheelchair during long walks to relieve you, or use an all-terrain wheelchair, etc. It finds that adults Angelman have quickly renounced an activity if he is asked too much effort or puts them in trouble. A certain passivity and denial of exit are sometimes not favouring the maintenance of motor skills and activities, but the Angelman Syndrome are, like everyone else, subject to normal aging and age-related diseases that sometimes slows down the activity.

Switching to adult institution intervenes later, around the age of 25 years, due to the lack of suitable places. This is a new step that requires a big effort for adaption, especially for those who discover the internship. The Angelman leave an educational universe to discover a new place to live with often a very different rhythm and the encounter of adults with disabilities of all ages. As institutions, they are very diverse, some without that of the occupational, while others are attentive to the persistent evolution of adult Angelman.

With the development of establishments catering for people with disorders of behaviour (autistic type), we see develop new types of adults supported, between education and creation of activities sticking to everyday life, where everyone can participate at his level to the proper functioning of the group. Probably a plus for the Angelman arriving to integrate these structures as we have seen in our study.

Thanks :

We thank the French and foreign families who participated in this survey, AFSA and the association Des Anges Pacôme, otherwise (DAPLA). And for the translation : Harrold Simpson.

References:

- (1) J Clayton-Smith, L Laan the syndrome d'Angelman: **review of clinical and genetic aspects** (2003)
- (2) A J Dagli and C A Williams: Syndrome of Angelman (1998 - last updated 2011)
- (3) L Laan, van denBoer: **Angelman Syndrome in Adulthood** (1996 - Netherlands)
- (4) J. Clayton-Smith: **Clinical research on Angelman syndrome in UK** : observations on 82 affected individuals (1993)
- (5) . Karine **PelcGuy Charles**, and Bernard **Dan**: Behavior and neuropsychiatric events in **Angelman syndrome**

The association **Syndrome Angelman France** aims to be a resource centre for families by offering such exchanges and a sharing of experiences in order to help better support the Angelman as well at home than in institutions and get a better taking into account of their disability in society :

www.syndromeangelman-france.org