French Association of
Costello & Cardio-Facio-Cutaneous

Le Petit Journal

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Research projects

ICS is 20 years old Dr Yann HERAULT & Tania SORG

COSMITO Dr Rodrigue ROSSIGNOL and Laëtitia DARD

Information

- Augmented Alternative Communication
- Parent carers

Testimonials

- of the Institut de la Clinique de Souris in Strasbourg
- of our families
- of professionals who follow our adolescents

Other associations

- of our families, or in the course of our meetings

Those who support us

- Coureurs Solidaires, Mascaret, Open Garden, Lyre & Elles

Family gatherings

2019: face-to-face, 2021 & 2022: distance learning

And always so rich and well followed

From our medical committee :

Discussion groups Eva TOUSSAINT

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The President's message

Hello to all,

This Journal in 2022 allows us to inform you of our actions to reinforce the knowledge on Costello and Cardio-Facio-Cutaneous syndromes in order to improve the care of the families.

But above all it is a link between us. And we are all missing one, whether we are a family, a member, or a non-member but someone who is sensitive to the daily life of families with a child, adolescent, young adult, or adult carrier of one of our two rare diseases. It is for this link that we publish this Petit Journal.

Several years have passed since No. 18... What's new?

COVID prevented us from meeting physically. In order to answer the many questions of our families, we, the members of the board and our faithful professionals, have done our utmost to hold webconferences. You can discover the topics covered. A big thank you to each "old" family who participates in order to share their experiences with the "new" families.

But we miss the physical contact. And we hope to be able to organise a gathering in 2023. We have to do it! And I thank each and every one of you in advance for your help, I think of Jean-Yves and Florence.

Children are growing up! The age distribution graph we present here is striking: Damien is 44 years old! Séréna 37, Laure our BIG Canadian 33; Tiphanie, Louise, Salomé, Tamarra, Bilal, Théo, Gilles, Jonathan, Romain, Laure-Anne from 24 to 30. Already!!!

Of course, we have a tender thought for each and every one of them, but we do not forget the questions of their parents for which we must try to provide answers. Because the passage to adulthood is often associated with a loss of reference points, the paediatric services are no longer available. The network of professionals is no longer the same, "Who to refer to? Who can point me to the right professional?

What about you? Surely you can help us: do you know a good physiotherapist, a dentist who takes the time to welcome the teenager and get him used to it... Yes ? Yes, maybe? So, write to us and we'll get in touch with our families, one of them is surely near you!

Have you been challenged by our 2022 greeting card? How did it look again, oh yes that's her on the left.

What is AAC? You will know because Anahita has taken the time to explain it to you. And if you have an experience, contact us to testify. It will be useful for our families who are wondering.

What about you? Do you feel like a carer? Are you a carer without support? Cristina devotes a chapter to this. Maybe you will find some useful information there. Not enough? Tell us, ask us your questions.

Research... 2023 will be the year of the preparation of a therapeutic trial, 15 years after François DUPUY asked Prof. Didier LACOMBE to create the conditions for researchers to work on the Costello syndrome: the creation of a COSTELLO mouse. We are going to take up the story with the Strasbourg Mouse Clinic Institute (ICS), with the researcher Rodrigue Rossignol and the post-doctoral student Laetitia DARD who describe their COSMIT project "Mitochondrial medicine for RASopathies".

ICS, the Strasbourg Mouse Clinic Institute is 20 years old. And on 17 and 18 November, Yann HERAULT and Tania SORG invited us to their congress, which brought together 50 researchers and technicians to talk about our common history. We were impressed to see all these experts, passionate people who can devote their lives to identifying the cause of a malfunction, and how to reduce it. We tell you all about it.

Here you are, read about us, discover our families, and don't hesitate to come and see us.

Serge ARNOULET, your President

Life of the association: François DUPUY

Serge has just returned from Strasbourg where the 20th anniversary of the ICS was celebrated... the Mouse Clinical Institute.

Some time after the creation of this institute, its director, Yann Hérault, published an article in a magazine open to all, La Vie, describing how studies carried out on a mouse model carrying trisomy 21 had made enormous progress in the knowledge of this disease...

Why not try a cooperation to find out more about Costello... The gene has been described since September 2005! It is only afterwards that tests done on all children diagnosed with Costello syndrome showed differences in genetic abnormalities, with RAS as a common point, hence the generic name Rasopathies.

Since the beginning of the association, the idea of creating a mouse model had matured... As soon as the gene was identified, we started looking for a possible coincidence... And in 2007, after contact with the animal house of the Pellegrin University Hospital, in relation with Pr. Mariano Barbacid in Madrid, a specialist in cancer and therefore in the HRAS gene, then with Pr Dubus in Bordeaux, himself in contact with Madrid and Dr Radvani, another cancer specialist at the Institut Curie in Paris... the rare object was found!

When asked, Yann immediately agreed and the "*Costello*" mouse began its adventure in Strasbourg... The other syndromes are too diverse to consider satisfactory mouse models.

Thanks to this "Costello" model, many experiments have been carried out to better understand the consequences of these syndromes. Some of these experiments have guided and advanced the work of Rodrigue Rossignol and Laetitia Dard (PhD student) in the laboratory directed by Professor Didier Lacombe on mitochondria... A molecule for a therapeutic trial is currently being marketed.

Since its creation, a whole network of researchers has been formed around these RASopathies ... at the ICS in Strasbourg of course but also at the Robert Debré Hospital in Paris, at the Timone Hospital in Marseille, and also abroad in Madrid, Manchester,... The scientific community is international... (The genetic anomaly

was found in a Japanese laboratory on French material with an Italian student trainee!)

So far we have managed to keep in close contact with all these people who have always been very available and attentive, thank you to them.

Many other experiments remain to be done... We must be available to the various remarks and requests of the families. Particular questions are asked by parents... to be reported to these researchers in order to understand the mechanisms involved... Two weeks ago we were still talking to a family about their son's sight, hearing and dental problems...

The association has lived for 22 years with 2 presidents ... who will take over ...

There is of course the administrative aspect with a treasurer for membership, mailings, etc...

It is also necessary to facilitate meetings/discussions between families, to help share the experiences of the older ones with the younger ones.

This relationship with research needs to be monitored and diversified.

WE HAVE TO GO! Come and join us,

it is the survival of the association that is at stake!

François DUPUY

Several families have joined us since 2020

In the Paris region

- Smail and Saliha live in Argenteuil, they are the parents of Rayane, 5 years old, carrier of the CFC syndrome
- Julie and Sébastien live in Savigny-Le-Temple, they are the parents of Charlie, 2 years old, carrier of the Costello syndrome
- Qi and Thomas live in Cormeilles, they are the parents of Kiara, almost 2 years old, who has Costello syndrome

In Brittany

- Natidja lives near Rennes, she is the mother of Nayilat, almost 3 years old and a carrier of CFC syndrome

In Normandy

- Lucie lives near Rouen, she is the mother of Léon, 1 year old, carrier of the CFC syndrome

In the Centre-Val-de-Loire

- Mélodie lives near Chenonceau, mother of Eliot, 2 years old and carrier of the CFC syndrome

Pays de la Loire

- Jean-Louis and Moriliat live near Saint-Nazaire, parents of Louis-Dayo, 8 years old, carrier of the Costello syndrome

In Toulouse

- Yang and Simon live in Toulouse, they are the parents of Léon, 2 years old, carrier of the CFC syndrome

Alpes-Côtes d'Azur

– Marília and Louis live near Monaco, parents of Liam, 4 years old, with CFC syndrome

On the island of La Réunion

- Clarisse and Jean-Jacques live in La Possession, they are the parents of Elia, almost 4 years old, who has CFC syndrome

In England

- Virginie and Michel live near Teddington and are the parents of Sophie, 3 years old, who has Costello syndrome

In Belgium

- Réginas and Diane live near ANTWERP, parents of Noam, 5 years old, carrier of the CFC syndrome
- Kate lives in Gembloux and is the mother of Lea, a 4 year old Costello syndrome carrier.
- Aurélie lives in Liège and is the mother of Théo, aged 2, who has Costello syndrome.

In Morocco

- Zineb and Aziz live in Agadir, Morocco, and are the parents of Youssef, who was born a few months ago with Costello syndrome.
- Asmae lives in Morocco, mother of Mohamed Majd, 3 years old, Costello syndrome carrier

We will do everything we can to help them and to enable them to come to the 2023 gathering, to benefit from the feedback of other families, from the conference-exchanges, from the individual consultations with the doctors who have been following our children for years.

We are calling on "old" families to come forward to offer a **cousinade to welcome our new families**. Contact us and we will put you in touch. We will help you to organise it!

Families: don't forget that we can send you a list of all the families by department, *(see the breakdown by department a few pages further on)* so you could discover future friends close to home. Write to us!

New families. Our families 10

The association refers 123 families and 119 carriers of one of the two syndromes:

55 carriers of Costello syndrome,

64 carriers of the Cardio-Facio-Cutaneous syndrome.

The association is **22 years old** and the graph above shows the distribution of the number of carriers by age *(we don't always know)*

Nbr	S		CFC	C	Tot
Age	CFC	Tot			
1	1	1			
3	3	3			
4	2	2			
5	1	1	7	7	0
7	1	1			
8	1	1			
9	4	4			
10	1	1			
11	4	4	11	11	0
12	6	6			
13	3	3			
14	3	3			
15	4	4			
16	4	4	20	20	0
18	1	1			
19	1	1			
21	2	2			
22	4	4			
23	1	1			
24	1	1			
25	2	2	12	12	0
26	1	1			
27	1	1			
29	1	1			
Tot	53	53			

16 children are under 5 years old,

13 children from 5 to 8 years old,

28 pre-teens - teenagers aged 9 to 14,

28 young adults aged 15 to 21,

19 adults aged 22 to 26,

8 adults aged 29, 33 and 40

33 are civilly adult

Our association must find ways to help young children, adults in the making, and adults seeking a future.

The transition to adulthood is a subject we wish to advance from a medical and social point of view, but also to enable their integration into society.

A large majority in France

Nom R	Ancienne Région	Total
⊟	Auvergne-Rhône-Alpes	5
⊞	Auvergne	2
⊞	Rhône-Alpes	3
⊟	Bretagne	4
⊞	Bretagne	4
⊟	Centre-Val de Loire	2
⊞	Centre	2
⊟	Grand-Est	5
⊞	Alsace	1
⊞	Champagne-Ardenne	1
⊞	Lorraine	3
⊟	Hauts-de-France	3
⊞	Nord-Pas-de-Calais	3
⊟	Île-de-France	31
⊞	Île-de-France	31
⊟	Normandie	2
⊞	Basse-Normandie	1
⊞	Haute-Normandie	1
⊟	Nouvelle-Aquitaine	8
⊞	Aquitaine	4
⊞	Limousin	2
⊞	Poitou-Charentes	2
⊟	Occitanie	5
⊞	Languedoc-Roussillo	3
⊞	Midi-Pyrénées	2
⊟	Pays de la Loire	5
⊞	Pays de la Loire	5
⊟	Provence-Alpes-Côte d'A	11
⊞	Provence-Alpes-Côte	11
Total général		81

⊟ Guadeloupe	1
⊟ Guyane	1
⊟ La Réunion	1
⊟ Nouvelle-Calédonie	1
⊟ Polynésie Française	1

but also **33** worldwide,

hence the acquisition by the association of a tool for automatic translation of our documents

Pays	Total
⊞ ALGERIE	2
⊞ ALLEMAGNE	1
⊞ ANGLETERRE	1
⊞ ARGENTINE	1
⊞ BELGIQUE	4
⊞ CANADA	4
⊞ COLOMBIE	1
⊞ ESPAGNE	5
⊞ ITALIE	3
⊞ MAROC	1
⊞ PAYS-BAS	3
⊞ PORTUGAL	3
⊞ ST DOMINGU	1
⊞ SUISSE	1
⊞ TUNISIE	2
Total général	33

Research projects

The Strasbourg Clinic Institute is 20 years old!

Dr Yann HERAULT and Tania SORG, from the Strasbourg Clinic Institute *(see Petit Journal 2016)*

The Institut Clinique de la Souris was 20 years old and Serge Arnoulet from AFS-Costello-CFC was with us to blow out the candles.

The ICS was created in 2002 by Professor Pierre Chambon with the aim of providing a high-performance, high-throughput tool for the generation and functional study, phenotyping, of genetically modified mice. Over the years, the main missions of the ICS, as a service infrastructure, have become 1) To be a service platform for research combining the capacity to generate, on a large scale, targeted mutations in mice with comprehensive high-throughput phenotypic analysis of the mouse with the aim of facilitating the creation, analysis and use of the mouse model; 2) To maintain relevant expertise in both in-house research and development programmes to

support ICS activities in the field of mutagenesis, transgenesis, phenotyping, bioinformatics and data analysis; 3) To be a reference centre in the field of mouse functional genomics and translational research; 4) To promote the best training both internally and for users to ensure the most efficient procedure in respect of ethics and animal welfare.

The ICS is thus able to manage and carry out each of the key stages of a project, from the generation of genetically modified mice, reproducing the mutations encountered in the human clinic, to the characterisation of these models and a better understanding of physiopathological mechanisms, to preclinical studies to evaluate therapeutic treatments. During its 20 years, the ICS has contributed to the generation of more than 2500 models and the realisation of more than 1200 projects of phenotypic characterisation or preclinical studies. In addition, the ICS has joined the PHENOMIN initiative, winner of the national Health and Biology infrastructures within the framework of the investments for the future in 2011, as well as the European INFRAFRONTIER infrastructure and the international IMPC Phenotyping consortium initiative, to strengthen its position in functional genomics and translational research.

At this 20th anniversary event, we were able to appreciate and share with stakeholders from academic and private laboratories, as well as our partners, such as the Rare Disease Foundation and patient organisations, a set of discoveries that are of paramount importance for fundamental and biomedical research and therapeutic innovation. We reviewed the latest developments in various fields, from mouse genetics to its use in understanding rare diseases, neurodevelopmental and degenerative diseases, and in immunology, inflammation and infection, and ended with a tour of our Institute.

For more than 20 years, the ICS has been contributing to research on rare diseases. In this context, our interaction with the AFS-Costello-CFC is exemplary and unique. It underlines how we can develop a model to push

research and observe new discoveries on Costello disease and the emergence of therapeutic leads.

Serge told us our common story for the last 10 years: the meeting with François, Didier and Rodrigue, Yann and Tania, and of course the meetings with the families in Bordeaux. Starting with the generation of what we call at the ICS "the Costello mouse", and then funding from the French National Research Agency to characterise this mouse, which has strong similarities with the patients, we have been able to begin to understand the mechanisms involved in the disease and to start a therapeutic study aimed at correcting certain symptoms. We hope that these trials will be successful. However, we must remain cautious, even though we understand the urgency, and wait for further advances in our knowledge of the disease to ensure the success of future treatments and to facilitate the lives of patients and their families.

Dr Yann HERAULT and Tania SORG

The COSMITO project

Mitochondrial medicine for RASopathies.

Laetitia Dard, Nivea DIAS AMOEDO, Didier Lacombe and Rodrigue ROSSIGNOL (U1211 INSERM, Bordeaux).

The RASopathies[1] are rare genetic diseases that share a common feature of hyperactivation of the RAS/MAPK signalling pathway due to mutations in several genes (HRAS, KRAS, NRAS, SHP2, Grb2, SOS1, NF1, BRAF, RAF1, MEK and ERK). The RAS/MAPK pathway plays a major role in many cellular and developmental processes such as cell growth, cell division, metabolism and organism development. Costello syndrome (CS) is the best described RASopathy caused by mutations in the HRAS gene, the main symptoms of which are growth retardation, dysmorphia, delayed cognitive development, skin alterations and a predisposition to certain cancers. To date, there is no curative treatment for HRASopathies. A central feature of Costello Syndrome (CS) and other RASopathies is **hypertrophic cardiomyopathy**

(HCM)[2] **,** but the molecular mechanisms linking HRAS activation to this cardiac dysfunction remain unknown. However, cardiac involvement is a major determinant of the prognosis of HC, raising the need to identify the molecular causes of HCM and to propose appropriate therapeutic strategies. An estimated 100,000 children are born with hypertrophic cardiomyopathy (HCM) each year worldwide among individuals with RASopathy. The clinical management of cardiomyopathy associated with RASopathies remains a challenge for cardiologists and there are no drugs on the market to treat the causes of HCM. According to the American Heart Association, "**alterations in mitochondrial function are increasingly recognised as a contributing factor to myocardial infarction in patients with cardiomyopathy**. In line with this statement, we have recently shown the existence of early alterations in cardiac bioenergetics responsible for the development of HCM[3] . Recent work also shows that **mitochondrial inhibition is a common early mechanism for this condition, regardless of its genetic origin** [4]

To date, there is no effective treatment to prevent, slow down and treat HCM in RASopathies and its more common forms. The only option for the clinical management of heart disease in RASopathies is symptomatic treatment of HCM with limited efficacy and safety[1] . First-line treatments focused on reducing the symptoms of HCM are offered to the majority of patients, but none of these treatments target the cause of HCM and the pathophysiological mechanisms of RASopathies. General guidelines for the medical treatment of HCM include the use of drugs such as beta-blockers, dysopiramide and L-type calcium channel blockers to suppress symptoms and the degree of left ventricular outflow tract obstruction (LVOTO)[3] . This treatment remains poorly effective and many patients do not respond to

[1] *Endocr Rev 39: 676-700*
[2] *Int Med Care, 2019 doi: 10.15761/IMC.1000134*
[3] *Dard L. et al. J. Clin. Invest 2022 in press*
[4] *Circulation. 2021;144:1714-1731.*

beta-blockers. Surgery (heart transplantation) is then proposed for those who do not respond to medical treatment

Recent work shows that regardless of the genetic cause of HCM, **mitochondrial inhibition (bioenergetics deficiency) is a common early mechanism in this pathology[4] .** Mitochondrial turnover is a process that can be driven by drug approaches, and pharmacological activators of AMPK or PGC1α prevent the development of HCM in preclinical studies. However, such an approach has not yet been considered for the preventive treatment of HCM in RASopathies and there are no licensed drugs to activate these pathways.

Faced with this limitation in existing therapies, we have developed a specific molecular therapy called 'Belithine' (patented) which prevents or reduces the development of HCM, by targeting a pathophysiological mechanism of RASopathies: the alteration of mitochondria. Our INSERM 1211 unit and the medical genetics department (Pr. D. Lacombe) are specialised in the study of this disease and have been working with the French Costello Syndrome Association and CFC since its creation in the Nouvelle-Aquitaine region. We have focused our work on **hypertrophic cardiomyopathy,** which is a poor prognostic factor

Our study of the molecular mechanisms responsible for HCM in Costello syndrome revealed an inhibition of mitochondrial function as a result of the genetic defect (HRAS mutation). Our therapeutic strategy of 'mitochondrial renewal' by Belithin thus appears to be consistent with the molecular pathophysiology of HCM in RASopathies, but also in common forms of varied genetic origin. Furthermore, HCM is a progressive disease and correction of early bioenergetic defects could slow down the heart disease, despite an inevitable hereditary genetic determinism. However, our development strategy for Belithin will initially focus on the treatment of heart disease in RASopathies. The proposed drug combines two synergistic active ingredients aimed at renewing and enhancing mitochondria by acting at three complementary levels: mitochondrial biogenesis, supply of energy

substrates and activation of mitochondrial quality control. These three effects result in the restoration of mitochondrial function and the preservation of cardiac function

We thus propose a non-toxic and innovative approach to metabolic treatments based on the understanding of the molecular mechanism responsible for HCM in Costello syndrome.

Laétitia DARD and Rodrigue ROSSIGNOL

Augmented Alternative Communication. What is AAC?

C of communication .

A fundamental human right.

Communication allows us to develop and find our place in society. We communicate everywhere and all the time (asking, giving our opinion, expressing our feelings, telling stories, etc.)

A of Alternative .

Another way, another way.

An alternative communication is a communication that takes place in a way other than through speech because speech is absent for whatever reason (through signs, writing, communication boards, applications on tablets, podd* ....)

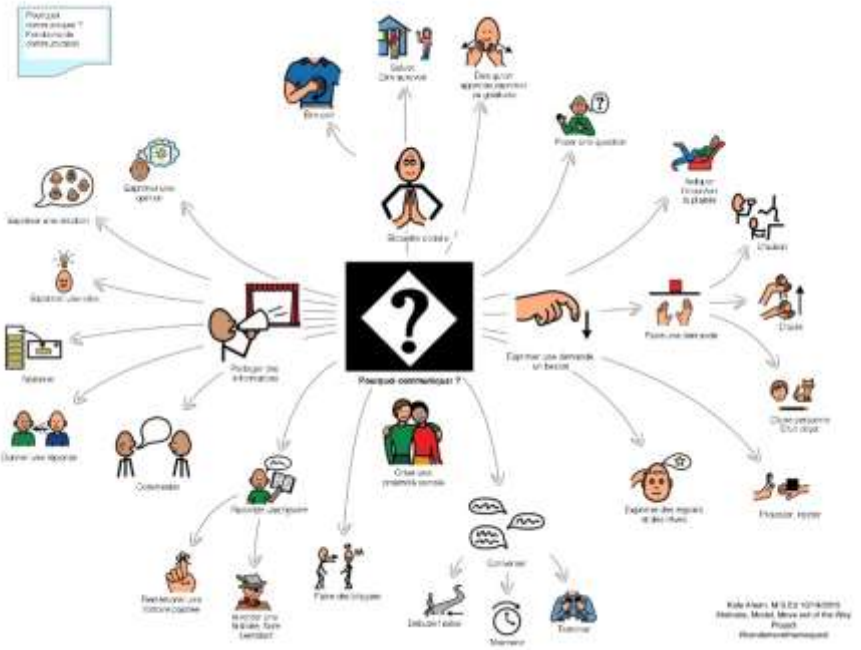
A of Increased or Improved.

A supplement, an improvement.

Augmented communication is communication that uses speech and other means that complement it (signs, gestures, writing, communication boards, tablet applications, podd*...).

** A PODD is a book of pictograms.*

Imagine you can't talk, how do you do all these things?



To make friends and not feel lonely anymore? How do you express your needs? At the doctor's surgery, for example? .... There are many situations in everyday life where we need to communicate. So we need to find a way to do this.

These means exist and are available to everyone, whatever their age or situation. Whatever the disability, it does not prevent us from looking for a means of communication to improve the person's quality of life. There are many tools available, it is necessary to find the one or ones that best suit the person. There is not one solution for everyone. A person may even start with one tool and continue with another or several others.

AAC tools are often classified by their level of technology. Those that do not require any technology, such as Makaton, which uses sign language and pictograms, or **PODD**, which is a book of pictograms. And tools that use technology such as tablet applications, TDsnap, P2go, Avaz, optically controlled tablets...

I am the mother of Ulysses, a 20 year old man with Costello syndrome. Ulysses was introduced to signs at the age of 2. It was in signs that he was able to express his desire to listen to music, it was in signs that he was able to express that he was thinking about his father or that he was happy. It is thanks to signs that he became clean .... signs accompanied him for a long time and still do. I think they are a very good AAC tool for the youngest children and a complement to another tool. But signs have the disadvantage of not being understood by everyone. We, at the time in France and then in Mexico, had no access to anything else. Ulysses was introduced to Makaton which uses signs and pictograms. The pictograms allowed us to create (life) books which, like a diary, allow us to tell our daily lives, what we do and how we feel. The life book is also a tool of AAC since it allows to initiate the conversation, to express oneself and to maintain social

links. Finally, 5 years ago when the first iPad communication application became available in French, we adopted it so that Ulysses could take the path of the most complete and autonomous communication possible. Ulysses is still working on it every day. This work allows him to improve his language and to make progress in the acquisition of reading and writing (literacy).

AAC is a very long way.

It takes patience and perseverance, you have to assume the person's potential and not the other way round, you have to invest yourself in it (I mean us, the parents, the siblings, the relatives), it's not only the speech therapist's job. But it's worth it because it allows the person we love and who needs our help to move forward, to blossom, and to enjoy all their rights as a human being.

Anahita, mother of Ulysses 21 years old

Links to find out more

AssistiveWare: many articles to start or deepen your knowledge
https://www.assistiveware.com/fr/apprendre-caa

CAApables: including infographics, info on myths and PODD starter pack
http://www.caapables.fr/

HappyCAA: more for families, testimonials and articles https://happycap-foundation.fr/caa/

CAA pratik: including nice videos and infographics https://caapratik.com/

Keep smiling: lots of books on AAC, among other topics
https://www.youtube.com/channel/UCrtbeVz6oOKealwyFEf9GXA

A plusieurs voix: research articles translated into French
https://www.aplusieursvoix.com/

CAAusette: https://www.caausette.com/

French ISAAC: https://www.isaac-fr.org

Anahita, mother of Ulysses 21 years old

Parent carers, don't be alone!

*** The financial aspect :**

First and foremost, if you are a new parent of a child with multiple disabilities, you must apply to the CPAM and the MDPH for *financial support and, above all, to obtain recognition of your child's pathology, which will allow you to benefit from appropriate social security coverage.*

*** A unique path for my child**

The association's experience has shown that there is no homogeneous pathway. There are as many paths as there are patients. It is up to you to knit your own with the tools that exist according to your child's needs. But don't be alone. Ask for help, knock on doors.

Depending on the age of your child, there are experienced people whose role is to provide guidance and support in administrative procedures and in the choice of educational or medical/social care.

What support for my disabled child? CAF website :

- CAF (education allowance for disabled children, AEEH)
- The PMI (Protection maternelle infantile) of your department can help you.
- The MDPH (recognition of 100% coverage)
- Home help association (ask for the list approved by the MDPH of your department)
- The CAMPS (Centre d'action médico-social précoce)
- The IME medical-educational institute
- The social worker of the above mentioned institutions
- The psychologist of the neurology department or of the institutions mentioned above

This list is not exhaustive and it does not cover all situations, but it does give some ideas to explore.

*** Respite for parents**

You should know that IMEs, follow-up care centres and specialised establishments exist to take our children for short periods of time, these are called "respite stays". This is done in conjunction with a social worker from the above-mentioned institutions. A joint project is drawn up which can be long term or ad hoc depending on the situation. It is a tailor-made project.

- Good news for carers:

- There are now specific cures for family carers, which are paid for by the MDPH. A request must be made to the treating physician and the MDPH.
- In France, various health resorts offer dedicated stays, for example
  - the aid module at the Lamalou-les-Bains thermal baths
  - the aid module at the Saint-Honoré-les-Bains thermal baths
  - the module helping at the thermal baths of Mont-Dore
  - a mini thermal cure at the thermal baths of La Léchère-les-Bains
  - a "Help for carers" stay at the thermal baths of Néris-les-Bains
  - a "Quiet" stay at the thermal baths of Bagnères-de-Bigorre
  - Special cure for caregivers, in Hendaye (64)

The programme of these stays includes various **thermal water-based** treatments, meetings with professionals, discussion workshops and the initiation or resumption of an adapted sporting activity.

It's about taking care of yourself before you burn out. Taking care of a child with multiple disabilities on a daily basis requires so much that it has repercussions on your own health. You have to learn to pass the baton, which does not mean giving up, but taking care of yourself so that you can remain effective and last in the long term.

Cristina AQUABA mother of Neil, 13

Memories: an extract from Le Petit Journal from 2010

Do you like memories? I, Le Petit Journal, love to go back to my old issues to see our children, who have grown up a lot. So today, I'm sharing with you an extract from the 2010 issue about the rally. **Do you like it? Let us know.**

The May 2009 rally

We end up with 61 families and 5 educators or other contacts, scattered all over the world! For a newspaper I can be proud...I am translated in 5 languages !

In May in Gradignan, 6 of these new families met up with 6 of the older ones. They have already told you a lot about it in a special report, each one of them expressed what they had experienced there... but with a lot of restraint.... I, who am only a diary, something that is left lying around and that we are not suspicious of, was able to see reddened eyes, parents who were a little knocked out by what they had just heard from other parents during the round tables or from the few doctors present for the consultations, but also a dining room or a courtyard and a park full of good

humour and, dare we say it, happiness for some! I can't resist showing you a few photos taken by some and others and compiled by Jérôme!
During this event many people were there to organise and provide logistics, and to look after and/or supervise activities for all the children. There were many who had not drawn or told stories or pushed a pram around the park for a long time... Thank you to all of them.

I would like to share with you a special thought for Christian Dubois, this bearded gentleman who helped children to express themselves by making objects and characters out of cardboard and polystyrene...
Christian left us suddenly in October... He who seemed so taciturn had told us how much that day with those children had upset him. He had rediscovered the taste and meaning of his work as an educator and of many things...
Goodbye and thank you Christian!
There is so much to say here too that I don't hesitate to show you pictures to illustrate it all! ....

Testimonies of the families

The PNDS: a family's testimony

When I first read the PNDS (National Protocol for the Diagnosis and Care of Rasopathies, NOONAN and CFC), Neil, my son was 9 years old. I had just been diagnosed, the genetic service that followed him had just identified the name of his disease, Cardio-Facio-Cutaneous Syndrome (CFC).

As I read through this booklet (PNDS) my eyes widened, I saw the care pathway pass before me; the different areas of his medical care; almost all the problems of our experience from the point of view of symptomatic manifestations; examinations; assessments and decisions taken. A part of his life in a nutshell.

This support is very important, it can prepare you to some extent for what you are likely to face. It's a great thing to have. I encourage parents of children with CFC syndrome to read it, not only does it answer many questions but it also gives many tools to guide the health and care pathway.

Crisina AQUABA mother of Neil, 13

The Costello NDP: in 2023 we will update it

The Costello PNDS is old, it was released by the HAS (Haute Autorité de Santé) in July 2012. Since then, our families have noticed that certain topics have not been addressed, even though they are critical to the daily lives of children, adolescents and young adults.

Doctor Sabine SIGAUDY, who has been following our families since the creation of the association, and is present at each of our family and professional gatherings, has proposed that we coordinate the actions to update the Costello PNDS.

We will ask each family to work on this using the structure of the Noonan and CFC Rasopathies NDP.

Meeting an angel

Our little girl, Melissa, had Costello syndrome. Like many parents, we were already making up our minds while waiting for her to come, about who our child would be. The first four months after her birth were difficult. Between doubts, symptoms and then the diagnosis.... It was a shock. Like everyone else, we rushed to the internet to find out more and understand.... What a mistake...we became afraid. We had a child with Costello syndrome....

And then, looking at this little flea full of life, marvelling at everything, we opened our eyes. She showed us that she wasn't her syndrome, that she didn't define herself just like that, pigeonholed by this particularity that Nature had decided for her.

Yes, there were difficult times.... But that was nothing compared to the happiness we shared with her.

There are no words strong enough to describe who she was. She gave out her tenderness without counting the cost, she gave out so much light that she lit up the darkness. She smiled constantly and fought every day to make progress as if she knew deep down that it was important.

We went along with her, not caring what anyone said, just following our instincts, knowing in our hearts that she was simply amazing.

She made the medical profession lie so many times, she made us lift mountains. How can you imagine that such a little girl could give off so much strength.

Our little angel passed away recently. Every moment we think of her, of all that we have lived through and her syndrome, which at the beginning was

our greatest fear, has disappeared day after day, leaving only such an extraordinary child, our child. We are so proud to have been her parents.

Thomas and Vanessa

Angelica's struggle

Angelica was born on 02 December 2011, she is now 9 years old but who would have imagined the hard road ahead? Who would have imagined that we would live a fight without respite? Who would have imagined that we would have the strength to face so many obstacles?

I, "Stephanie" the mother, and my husband, "José Luis", were completely happy to see the family grow. We already had a daughter "Carolina" who was then 6 years old and a boy "Joaquim" who would soon be 4 years old. Everyone was happy to see Angelica soon.

When the day arrived, we had no idea that this birth would change our lives... She arrived 1 month early but with a good birth weight of 3.680 kg., the delivery went well and everyone was anxious for us to get home soon. This happiness soon became a nightmare with worries, doubts but no answers. According to the doctors it was just a bad start, my baby was perfectly normal but the feeding difficulties were severe, she wasn't gaining weight and was regurgitating a lot, she didn't even have the strength to suckle. We spent our time in hospital. I could see that there was a problem. Angelica had a small tongue that often came out of her mouth, she was always very bent over and made little movements, the reflexes that I had known with her sister and brother were absent in Angelica. The delay in her development was becoming more and more noticeable. It was not until she was 6 months old that I was suddenly told that my daughter had a problem, given her catastrophic growth curve and her reactions. After a brain MRI, despite my doubts and questions to the professionals until now, I had been told that I had a perfect baby and that there was nothing to worry about, I was now told that my daughter might never sit up on her own one day and that we didn't know if she would be able to understand anything... I will

never forget that day, the announcement marked me deeply, I felt that there was a problem but I was far from imagining something so serious... The hospitalizations were multiplying and were long, Angelica was fragile. Her first 4 Christmases and her first 3 Easter were also spent in hospital. Finally, it was a feeding tube that was put in when she was 9 months old and that she would keep for 3 years that would gradually give her back a little strength. In addition to all this, we also discovered that she suffered from epilepsy and her sleep is very disturbed. We always had to repeat everything to the professionals, to the exams and to the people around us... it was very hard to explain to them because we ourselves didn't really have any explanations.

For the first few years we were afraid of losing her, we were lost, there wasn't even a name for her condition. After the announcement, I felt like everything was falling apart, I cried for several months, I isolated myself, why did this happen to us, as the months went by I felt a sense of injustice "did we deserve this?" and then I got angry... finally it was my older children who gave me the courage to pick up my head while I was spending all my time with Angelica, they were always very brave and understanding. I decided to stop working because I was too stressed by this situation and I said to myself STOP now we are going to do everything so that Angelica becomes as autonomous as possible, we are going to help her as much as possible to put all the chances on her side to live the happiest life possible. I think it is the wish of every parent "to see their child blossom and succeed in life" and we also want this for our children, our three children, and if Angelica needs a little more help for that, we are there, just as every parent responds when one of their children needs it, that's what life is all about: uniting and helping each other.

We quickly realised that this was not easy because the world in which most of us live is not yet ready to live with atypical people. Too many situations show us that when you are different you are quickly set apart.

Angelica has had to fight for her right to attend a mainstream school and not long ago she was in a shameful situation because her AESH did not change her nappy. However, Angelica has managed to find a place for herself in the school, the children are adorable with her and this relationship is good in both directions and this is also why we are fighting, to change mentalities, so that there is no longer any fear of approaching someone who is different, so that solidarity and mutual aid are meaningful.

Angelica attends school for 6 hours a week in CE1 with an AESH, she does not do the same work as the others but does fun things with her new AESH and shares with her class times of choir for example or with groups of two or three classmates does a time of socialization, she is blossoming thus. She is followed by a SESSAD for psychomotricity care, an educator, snoezelen time and by a physiotherapist in private practice. We have created an association for Angelica "AGISSONS POUR LE COMBAT D'ANGELICA!" in order to be able to finance intensive care abroad with the aim of improving her motor and cognitive skills and to improve her daily life to make her more autonomous and for her well-being. We inform ourselves about all the methods that exist in order to enrich ourselves to be able to help Angelica like the UPBRAINING method for example.

When we created the association, we did not know Angelica's syndrome, we learned that she had Cardio Facio Cutaneous syndrome on October 24, 2019, that's barely 1 year ago. This is how we were able to meet and exchange with the French Association of COSTELLO and Cardio-Facio-Cutaneous Syndromes and put us in touch with families who are in a similar situation and it feels good.

Angelica has come a long way but at her own pace she is moving forward and is happy, she gives us incredible strength, we love her more than anything. The future frightens us but we live in the present and fight every day so that her rights are respected and that she does not suffer injustice. We were told that "she might never sit up on her own one day" and well today she can move on her buttocks or with a little 4-legged friend, she

stands up on her knees. She doesn't speak yet but we are starting to communicate with pictures. She eats everything mixed but no longer has a feeding tube. With patience, extraordinary courage and an enormous joy of life Angelica is building her life. To all these rare pearls "BRAVO!", we have a lot to learn with these people and they deserve respect.

Stephanie and José Luis

Thirteen years of war

Testimony received in July 2020

I am Cristina AQUABA, mother of Neil, 13 years old, carrier of CFC syndrome, the disease was identified last year. We found out about the association right away and had the opportunity to participate in the 2019 gathering.

The members: parents; friends; relatives of the CFC & Costello association make up an authentic universe. Mutual aid and solidarity characterise the personality of the association whose members are united by a common cause.

My isolation has been broken, the network shares experiences; kindness and information make all the difference.

Since then, the daily struggle has been transformed because the association has already cleared many paths, nothing will be the same as before the meeting of these women and men who work in the shadows to help families.

Thirteen years of "war" have passed in the complexity of Neil's care: epilepsy, addiction, intellectual disability; behavioural disorders ...

I am now looking for a boarding school for my son to support me in his life.

Cristina AQUABA mother of Neil, 13

The Moulerens Gathering 2019, by Jean-Yves Faberon, Noumea.

At the meeting at the Château de Moulerens, near Bordeaux, we were the participants from the furthest place, the antipodes: Nouméa, in New Caledonia, in the South Pacific. We, that is to say the parents of David, 15 years old, CFC. We had never been to such a meeting, even though what has always concerned us is the lack of adapted structures in our country. We are lucky enough to have always had the annual visit of Professor Lacombe or someone from his team, and that is how we joined the association from the start. But as for taking part in its events, this imposes a long journey (difficult for the adults...so let's not talk about it, for our children) as well as very important expenses. So our situation with regard to David is one of isolation which often leaves us helpless. The kindergarten era, in retrospect, was the golden age, since at the beginning the toddlers are almost all at the same stage. But soon the gap widens. Primary school was a disastrous first experience for David, fortunately followed by a CLIS in another school where our son seemed to be as good as possible, so much so that we did everything to keep him there for as many years as possible. We were afraid to enrol him in an IME, looking for a school with a ULIS. We tried...but it was immediately catastrophic and we had to give up. There was no other option but to go to the IME with a heavy heart. David is still there. The establishment, materially, is not unpleasant, it is sunny, airy. David, most of the time, is there with a smile. But we are more than dubious about the possible development in a structure that takes in the diversity of disabled children. At sixteen today David, who does not speak well, cannot read or write. He is followed by the round of specialist doctors that all the members of the association frequent like us (with the added problem of his scoliosis and the -difficult- wearing of a corset).

When Serge encouraged us this year to come to the Moulerens Gathering, specifying the aspects of financial support that were of prime importance (and it so happened that I had planned to be present in May in mainland France, which was a good thing), we decided to come, both to

meet families in our situation for the first time and to consult the whole range of doctors available to us.

I will speak quickly about the medical consultations. I can only thank all the staff who are so devoted to our anxieties; some (not all) force our admiration. What they brought to us is important because they confirmed what the different doctors in Noumea had each told us. This guarantees us the quality of these. It confirms the pathologies of our child.

I must say that our great satisfaction in coming to the Moulerens gathering was the immense interest in meeting all this community, all these children: we were finally finding all these members of this kind of unexpected family that we form. Djamila Calin (who came to almost all the previous meetings) told me: I explained to my son Riwan that we are going to find our cousins! She is right, because seeing all these children, they do indeed look like family. And yet! If there is an important contribution to be made from this gathering, it is also to see how different these children are from one another. We all have to put things into perspective. For our David, we were deeply in awe of, for example, Jonathan's perfect expression, of some children's ability to communicate by mobile phone: David is far from it; but let's not complain, as we have discovered that a meal can be a terrible ordeal when the child cannot swallow.

Of course, we asked most of the parents about the structure to which they entrust their children. We were surprised to find that the answer was most often: to the EMI! Yes, but tailor-made IMEs, in towns where there are several. Some parents (from Aix) told us about trips to Paris organised by the establishment... Moreover, it is not necessarily a question of stigmatising overseas: Gina Madeleine, who came from Guyana with her little Marc-Isaac, told me how satisfied she was with the IME where her son is in Cayenne, with a teacher she described as wonderful.

The meetings organised for groups of parents were of great interest: everyone, like us, could see that we should not feel alone. We were pleased to meet friends from other horizons: from Portugal, from Quebec... Our

meals, taken together, were a strong contribution to this interest in dialogue, all of us.

The Moulerens gathering was an admirable meeting of so many sorrows and even tears in a moving set of empathies, the coming together of so many different people from all over the world, united, however, by similar vital concerns. The siblings of these CFC Costello children coming together to play 'collectively': how heart-warming!

It must be said that we had sunshine, also in the sky, and that the castle of Moulerens is really a marvellous site: space, nature, horses, shaded paths...

And then, what we wouldn't have experienced all these advantages without: the incomparable welcome and know-how of Serge, as tireless as his collaborators, always kind and even-tempered as they had to manage a real multi-faceted undertaking: the welcoming of one another, the installation in the accommodation, the meals, the financial and medical aspects, the fun aspects: what they have achieved is, in short, incredible!

From the bottom of our hearts we say a very big THANK YOU.

Noumea, November 2019, Jean-Yves Faberon, Noumea.

Laure-Anne, 30 years old, drug-resistant epileptic ?

Today my daughter is 30 years old, so this is an opportunity to look back on all those years of pain and difficulties but also of shared joys.

Since her birth, she has had enough problems to be hospitalised far from me, since then the medical environment has been part of our daily life, visits to specialists, hospitals, x-rays, examinations, ... nothing very pleasant in a family life. The differences that we noticed led me to contact a CAMPS, a difficult meeting because we were beginning to talk about disability, she was already 6 years old. It was also the time when we had to think about finding day care that would suit her situation, because since I had resumed work, she had been with a childminder who helped us and surrounded us. Finding an establishment that corresponds to our expectations is a long and very difficult journey because the confrontation with disability is particularly painful, we will again experience these difficulties between the "child" and "adult" environments, but here the search for a day care establishment is even more difficult and complicated because few of the MAS visited correspond to what we want for our child.

Her health has always been difficult, first of all huge eating problems, a constant reflux leading to a gastrostomy (which we quickly regretted) and a permanent refusal to eat regularly (she agreed to eat properly when she entered the second establishment she attended, i.e. at the age of 14), a scoliosis which became more important over the years, A diagnosis of arthrodesis was made, a major operation that we took the responsibility of not having carried out and finally, at the age of 12, the appearance of epilepsy which resists all existing treatments. We are waiting for her to be eligible for medical cannabis but without much hope of improvement. Today, her epilepsy is getting worse and is becoming difficult to manage for her and for us.

Everything is very complicated with a child like our daughter, ordinary care quickly becomes a challenge (especially dental care as nothing

is designed for her), maintaining her professional activity and pursuing a "career", outings (her baby-sitter is much younger than she is), family relationships that become more complex, holidays (we have to find homes or conditions that fit our constraints), daily life (after our working days we start again once we have been at home for 27 years), sleep which is rather complicated since her birth, both ours and hers, and illness. It is not always easy in these conditions to maintain a harmonious link with our child and those around us.

Our luck and happiness is that our daughter is, in spite of all the difficulties and constraints, a joyful and extremely kind child, and she is also very cuddly, which delights me every day. We are also lucky to have gone through these difficulties together, which is not always the case.

Annick VALLET

Raphaël, CFC, 23 years old, an "old family

My name is Raphaël, CFC, 23 years old. The former members of the association certainly remember me...

After the IME, I entered an occupational home not far from home. I was very lucky, I did a 15-day placement there and within a month my parents received confirmation that I was accepted, whereas it usually takes much longer, weeks, months. I took advantage of someone leaving in the summer of 2021.

So I am a day student, a city shuttle picks me up in the morning around 8:30 and brings me home around 4:30, 5 days a week. I have 5 weeks holiday a year, including 3 weeks when the home is closed for the summer in August.

At the home, I found some friends from the IME, fortunately because the population is older than me. We have many and varied activities, no respite:

- sports (judo, archery, introduction to rugby, introduction to sailing),
- looking after the animals at the educational farm next to the hostel,
- cinema (but I rarely have the patience to sit through the film...),
- library,
- relaxation,
- dance,
- music,
- plastic arts,
- petanque,
- shopping for cakes, etc.

I'm always mischievous, I make my teachers see it, but my parents don't know anything about it: what happens in the home stays in the home.

At home, it's simple, I don't do anything, much to the parents' despair! I spend my time on my games console, I love Mario, and on my tablet. I'm pretty good at it and always amaze my parents when they see me surfing with dexterity.

Health-wise, not so good. Epileptic seizures, often mild but very tiring. My eyesight is slowly but surely deteriorating, as is my hearing. I'll soon be wearing hearing aids.
Apart from that, I'm fine. I still have problems expressing myself and it annoys me when people don't understand me. I have a good appetite and eat almost everything. I'm 1.63m tall, and weigh 46 kilos wet, so I'm pretty small.

The holidays? Always with my parents, they don't leave me alone. Generally in the mountains, I like to hike in the peace and quiet, at altitude, in the fresh air!

For administrative matters (MDPH, AH), it is my parents who take care of them; they have been granted family capacity by a judge since I was 18.

I hope to see you all again at a future meeting, but in the meantime, don't hesitate to contact me (or rather my parents, who are the secretariat) if you need more information.

See you soon, friends.

Raphaël, 23 years old

Isorée, family member, caregiver at every gathering

I was asked to write about Costello and given carte blanche. What could I possibly write about Costello...

For as long as I can remember, Costello Syndrome has been with us. It came into our family by touching a little angel, who has since been called back to heaven. When I was little it was something easy, playing, drawing and learning to count, I don't even remember seeing a real difference between her and me. She was a child like me who I laughed with, we played at being mums and dreamed about our future life. Unfortunately, with this syndrome, future life is more of a daily struggle and I understood this thanks to the association. It was created to allow families to get together, parents to support each other, brothers and sisters to talk and children like me to think and understand.

Over the years, my relationship with the children has changed a lot. At the beginning I was the one who had a temper tantrum because she didn't see why Diego couldn't play football with us. Then I became a volunteer, at the first meeting I was 15 years old. In just three days I left there crying and feeling things that still affect me today.

I came in thinking I knew everything, saying to myself "I know what a Costello child is" but not at all: the first thing I learned was that each child is unique and that I should adapt to each one. Because, yes, like all children, they have their own characters, their desires, their tantrums and between the one who wants to be left to play on his own and the one who must not be let go, trying to find his place is complicated. They also each have their own way of talking, of telling you that you're pissing them off and of saying thank you. So you have to have a lot of energy and be very attentive. When I go to the meetings, I do it for only three days and I come out totally drained. So I wonder how parents do it. How do they have so much energy, how do they keep fighting every day, how do they listen,

How do they manage to understand their language, how do they manage to never give up?

How I admire her parents whose courage, love, patience and fortitude are remarkable.

What strikes me most at the end of each meeting is that I am able to communicate better with them than with some people around me. The discovery of this form of communication has been revealing in my relationship with these endearing children who make me grow and mature.

I end by thanking all the incredible people I have met, who I support and to whom I send all the most positive vibes I can.

Isorée Dupuy.

Our first participation in a gathering

In 2019, it was our first participation in the family gathering. We both came with Léonce, leaving his dad at home with little brother Gaëtan. The arrival was eventful, I was very emotional and intimidated myself, and Léonce was scared in front of all the children who were gathered at that moment to go pony riding. She cried, cried, cried, and even blamed me for bringing her...She then refused to leave the room until the next morning. I was totally distraught, and I cried a lot too. Fortunately Sylvie consoled us and even lent us her car so that we could both go into town to eat.

In the morning everything was better. Léonce met up again with her great friend Elisa, whom she met in Mayenne in 2014 for the notube weaning. She also got to know Malie, Honorine, and a lot of other children... to the point that I didn't see her for 2 days! Incredibly, she was going to eat with the friends, whereas she has always demanded to sit next to me whenever there is a meal, and never eats if I don't eat myself... The miracle of

togetherness. She was doing her own thing, going on the bouncy castles...incredible!

For my part, I was very moved by the attentions of the volunteers, all the kindness shown to us, and I felt touched by all the families I met, and very proud to be part of this big family. The involvement of such concerned professionals was also very reassuring.

Since then Léonce has gone back to Ulis for her last year. She is doing rather well, even if at 11 years old you can feel adolescence coming on and the sunny, laughing little girl is turning into a sulky, grumpy pre-teen... we have started a growth hormone treatment which makes her tired, but we have the impression that it helps her tone up, so for the moment we are continuing. I often think of the families I've met and the richness of the moments shared, hoping there will be many more.

Bérangère, mother of Léonce 13 years old *today*

Louis, Yohan's big friend at the 2017 rally

Louis with his bicycle transformed into a draisienne.

Louis is a fan of motorbikes, like his father. So he has his helmet.

Louis is now 13 and a half years old and for those who remember, he still loves to dance. He has become a real big boy with a lot more confidence.

His character is now well marked, if you know what I mean. With his parents at least, because everyone who looks after Louis describes him as cheerful, smiling and full of energy. Which is absolutely true. A real ray of sunshine and well in his skin, we are told. Nothing could make us happier. As far as his intellect is concerned, Louis is evolving well but remains a little boy.

In terms of learning, he knows how to go to the toilet alone to urinate and he is starting to want to be alone to have a bowel movement. This is such a great victory for us! He has been suffering from enuresis for 2 years. Before that he was independent at night.

Louis is monitored and takes a hormone every night, which allows him to urinate much less at night and is safe.

He can speak a few words and continues to make efforts, counting to 4 and 10 in his own way. However, when he was 6 years old, his neurologist told us that he would probably never speak. So let's not forget that a human being is unique and that we should never lose hope.

Louis loves to ride his bike. He rides very well but without the pedals. For the past 3 years we have converted two ordinary bikes into a draisienne, by removing the entire pedal mechanism. This allows him to have a bike that fits him. This has enabled him to make a lot of progress in his motor skills. And a lot in terms of balance. He now runs as fast as we do. If not faster! His eyesight is not optimal, but we have trouble getting him to accept a full examination. That's why his eyesight is corrected, but only approximately.

His food allergies are still present but now that he can eat eggs it is easier. However, his favourite foods are still chocolate, biscuits, yoghurt and cheese. He still eats a balanced diet.

I could go on and on about Louis, telling you how well he is doing, how happy he is.

However, we have to keep an eye on his behaviour as he sometimes becomes angry. We think that the difficulties he has in expressing himself play an important role. The few words he masters are obviously not enough.

Yet we are so proud of him. I will never stop saying it, he is teaching us about life.

Caroline, mother of 13-year-old Louis (C.f.c)

Professionals testify

Psychiatrist: Lisa's progress at 26 years old *today*

I have known Lisa since 2017

At the beginning, she was in a certain communicative closure, a kind of hesitation to present herself to the other, to take place in the relationship, limited by her difficulties of locution and also by a great lack of confidence in her expressive means. Lisa struggled to assume the dimension of a differentiated and individualised subject in relation to others. She remained imprisoned in the traumas present in her medical and societal history, the narration of which was difficult. At first she existed in the relationship, notably through her mother's speech. Her emotions were overflowing, especially anger, and their regulation depended on the containing and reassuring presence of her family members.

Lisa has invested our sessions well and we have been able, she and I, to feed our common work of psychic elaboration in continuity. She is able to better take into account what is agitating her inside and to seek its resolution, a possible appeasement to be more available in the investment of her daily life.

Lisa finally states her discourse around her personal evolution, she is the narrating subject, committed to her desires and desires in relation to her existential horizon. She is both more aware of her fragilities and limits and more determined in her daily commitments.

Her speech is now taking on a new volume in the relationship with the other, clarifying her presence, her determination, and becoming a point of support for asserting herself. His emotional self-regulation capacities have grown, and Lisa is increasingly aware of her own functioning and vulnerability.

We continue to work together with interest, enabling him to make steady progress as a person who can finally think and find his place among others.

Dr MANLIO SCIOMMERI, psychiatrist

Psychoeducator: Lisa's progress 26 years old *today*

When Lisa asked me to write a testimonial about her for our Thursday meetings, I was at a loss. What can I say, as a professional psycho-educator, about this courageous and determined young woman who has chosen to return to school where she left off? So I thought I would tell you about our meeting. I knew absolutely nothing about CFC syndrome. I therefore started working with her based on her strong desire to return to school.

In my psycho-pedagogical practice, I rely on two principles that are essential to the treatment.

The first principle is that of cognitive educability, i.e. that we can all learn whatever our situation, age and difficulties. The second principle is that of taking into account the request of a subject who wishes to go beyond his current difficulties.

To listen to Lisa is to discover a subject who tries to find a place despite all the difficulties she encounters. Her life drive feeds and irrigates her work and allows her to overcome the physical and psychological obstacles that this syndrome imposes on her. Lisa is a window to hope. She makes her way and it is with a small voice that she very discreetly tells us "it is possible" even if it remains difficult for her and her family.

Taking the time to listen to her, to walk alongside her, means sharing with her her questions, her doubts, her hopes and for me it is a real lesson in professional life. The learning situation circulates between us in a real pleasure of sharing.

Véronique Foll, Educational Psychologist

During the lockdown, I promised myself that if all went well I would resume swimming lessons.

In September 2021, I went to my first swimming lesson with back pain from scoliosis and shoulder stiffness from 16 years of heart surgery.

I've already had courses with several instructors but without success.

Eliot was able to manage and adapt to my problems of visual impairment, slowness, pain and fears.

Today I feel like a fish in water.

Lisa

"I'm very happy with the progress made with Lisa. We started last September with the aim of learning to swim, and her progress is being made session by session, which is very motivating for the future.

Today Lisa moves around in a prone and supine position with the help of a fry and an external support. Determined to learn to swim, she will continue her aquatic quest.

Eliot Bannerot

Master swimmer and sports coach

Our families' associations

Let's take action for Angelica's fight!

We created it in the summer of 2019, we didn't know yet what syndrome Angelica had but we found out just a few months later, in October 2019.

We created this association because first of all we needed to challenge and make others react, we needed to say what we experience on a daily basis and to make our distress understood. We also needed help and to make people aware of rare diseases and to know how to accept them in today's society by living together.

Personally, I shut myself away more and more after Angelica's birth and thanks to the association, I was able to open up to others again, there is more socialisation and beautiful encounters, you feel less alone. It's not always physical encounters but even through social networks we have enriching exchanges.

Angelica is already 10 years old now, when we created the association it was also with the primary aim of being able to offer her the maximum of tools and to make her benefit from all kinds of therapies brought to our knowledge to make her evolve as well as possible. We have been looking so hard for methods, materials, etc. that can stimulate her abilities to the maximum that through exchanges with other parents whose children also had disabilities (because I believe that parents of atypical children are often those who are the most expert over time and that is why exchanging our experiences is very beneficial on several aspects) that we realized and it's a pity but that in France at the level of the methods that exist to help our children that is not or still too little known.

For example, we have heard of intensive therapy centres abroad, but these have a cost.

After a first experience in one of these centres, first in Spain at our own expense, we realised that there were many things to stimulate our children

and help them evolve even more and this is what led us to create the association because these therapies are expensive but it is necessary to make several stays (at least once a year for results) and without the creation of the association to collect funds for this purpose we would not have been able to return. Recently Angelica did a 4 week intensive course in Portugal (2 hours of physiotherapy, 1 hour of PADOVAN, 1 hour of oral therapy and communication work and 1 hour of occupational therapy with also work on sensitivities) and she made more progress during these 4 weeks than a whole year with her usual therapies here. Our only regret is that we didn't get to know the centres and their methods earlier, because the earlier we start, the more beneficial it is. In addition, at the end of the course we are shown how to continue certain exercises at home.

We therefore organise solidarity races, shows, raffles, etc. to raise funds to continue these intensive courses abroad.

To make our association known is also to make known the syndrome of which Angelica is reached "the Cardio-Facio-Cutaneous syndrome" and to be able thanks to our association to intervene in the schools, colleges to make known this rare disease and our difficulties with the daily newspaper. It is also to fight for our rights, that they are equal for all like the right to schooling in ordinary school and to make accept the difference in our society because too many obstacles are raised on our way and it is for that that our fight is permanent.

Finally, when we can, let us not hesitate to pass on our knowledge to help others in similar situations.

The association has made us known and we feel more supported even morally.

We admit that because of Covid the events we organise have become more complicated to put in place and financially it is more complicated too but we keep hoping. Of course an association takes time and there is a little administrative work which is not always easy but often it is worth the effort.

We have plans to create a space to stimulate our children in France but it is a project that requires a lot of personal and financial investment but we do not lose sight of this objective because we would like to be able to do more so that our children can progress to the maximum of their capacities and continue to fight for living together and not "on the side".

The Facebook page of the association is: combatangelica

And the association's e-mail address is: combat.angelica@hotmail.com

For more information, please contact us :)

Stéphanie Marques Gomes (Angelica's mother).

Thank you to the AFS C & CFC for allowing us to make our association known.

Kisses.

Stephanie and José Luis

Au Fil de Léana

The association Au Fil de Léana was created at the end of 2015 by Aurélie and Gaël Perrouault, the parents of Léana, a 12-year-old girl with Cardio Facio Cutaneous Syndrome.

The desire to create this association arose in order to support the financing of equipment and therapeutic care not financed by the Social Security or the Departmental House of Handicapped Persons (MDPH) for Léana and other children with disabilities. Other aims: to raise awareness of the difference and in particular of disability and to highlight the role of family carers.

The association's income is mainly made up of donations, memberships and the sale of merchandise at events.

One of the association's particularities is to participate in sporting events (Thoiry Wild Race, Disneyland Paris Half-Marathon, 10 km of Trappes) and to carry out awareness sessions on disability for certain groups.

In 2020, the health crisis put a stop to many of the association's actions, which were mainly carried out in person. In order to continue its vocation and its actions by finding an alternative to the health situation in 2021, the association has created a virtual solidarity race. I'm putting on my trainers for Léana, it's :

- a concept: participants register on a dedicated online platform and complete a sports challenge from a choice of 6 options
- A balance sheet: 620 participants around the world and nearly 4600 km completed to further the cause of disability and support the association.

The association is also animated by social networks. More than 1500 subscribers to the Facebook profile and 350 on Instagram follow the adventures and actions of Léana and the association on the networks.

Our ambition for 2022 is to continue to advance the cause of disability and difference.

You wish to follow the news of this association or to support it:

On Facebook: https://www.facebook.com/aufildeleana
On Instagram: au_fil_de_leana
HelloAsso : https://www.helloasso.com/associations/au-fil-de-leana
email : auaufildeleana@free.fr

Aurélie and Gaël

Gatherings 2019, 2021, 2022

The proposed discussion groups : Eva TOUSSAINT : the key word is dialogue

The meeting days offered by the association are always an opportunity for formal or informal, serious or lighter discussions... In this article, we are going to talk about the discussion/meeting groups offered during the meetings and led by a psychologist. The aim is to enable parents and young people to meet and exchange views on their personal and family concerns in their experience with the syndrome. The groups are open to all but, generally, once we have started there is no more coming and going.

Each year, the topics discussed are varied, they are not defined in advance and it is the participants who choose what they want to share. Even if, in the end, some subjects come up every time, such as the announcement, the repercussions of the syndrome, emotional life, etc. Participants come to ask others about their concerns or to share their experience. Our attitude is not to give too much advice but to let the members of the group find the answers among themselves. We are there as a facilitator and not to give a theoretical-practical presentation. The idea is to distil information and notions of clinical psychology if necessary only.

An informal exchange in a spirit of respect and listening to each other is quickly established. These discussion groups take the form of an exchange, a free discussion which often continues beyond the group time.
I really enjoy taking part in these specific times of exchange: meeting up with parents or young people, seeing the richness of the exchanges. The most difficult thing is to have to slow down, organise and contain the words that flow out.

Taking part in these meeting days in a more general way is often very tiring but very positive for me. The possibility of meeting, of talking to

each other at different times, in different forms, formal or more informal, in groups or individually, in the rhythm and temporality of each person, seems to me essential and to be continued.

Eva Toussaint,
Clinical Psychologist - Doctor of Psychology - Family Therapist,
Developmental Anomalies and Malformative Syndromes Reference Centre - CHU-Pellegrin, Bordeaux

Why a gathering of families and professionals over almost a week?

Every two years since its creation, our association organises a meeting where families and doctors get together. The objective is multiple:

- break the isolation,
- inform families,
- promote the expertise of professionals,
- And allow siblings to confide in psychologists and other siblings.

This three-day break in their daily lives allows dispersed and isolated families to discuss, among themselves and with experts, their difficulties and the results obtained, both on aspects of child rearing and on clinical and psychological care (parents, children, adolescents, young adults and also brothers and sisters).

As these are rare diseases, by participating in a gathering, families can identify the specific experiences of their children with disabilities. For the doctors, this unique opportunity to see several people with one of our two syndromes allows them to reinforce or modify their expertise, and thus to better advise the families. For several of them, they have been following our families for more than 20 years, making themselves available to help them! We are very grateful for their advice whenever we ask for it.

Helping us find a place: an idea of our needs

Château Moulerens is ideal, with a very large capacity where we are alone in a secure environment for almost a week. But for the last 3 years, the site has been reserved all year round by the CRS. For 2023 we have a window in August. We are looking for other sites, some families help us, thanks to Jean-Yves and Florence! Not easy, because our gatherings include individual consultations with several professionals from all over France, hence the need for easy "direct" access by plane or train.

We have a lead for 2023, a week to be arranged between 12 August and 25 August, but perhaps we will offer you a few days in the Allier for the Ascension holiday...

We need your help to find a venue for our next meetings. So, to find out what we need, here are those of the last two face-to-face gatherings.

Example of our needs: at the Château de Moulerens - 20 to 24 June 2017

- 19 families: 26 children and young people (aged 4 to 28), 33 parents
- 20 members of the medical team (doctors, researchers, psychologists, speech therapist)
- 30 volunteers covering about 100 interventions (drivers, helpers, interpreters, accompaniers for outdoor activities, animation in the playrooms for the whole duration of the event, meal service...)
- Almost 700 meals served
- Accommodation on 2 sites:
    - about 40 rooms
    - 10 consultation rooms
    - 2 play areas
    - 1 reception room, a place for exchanges
    - 1 conference room
    - 2 group rooms
    - 1 activity room
- Covered space for table tennis
- Activities :
    - Climbing
    - Table tennis
    - Music
    - Initiation and pony activities
- Festive times (aperitifs, theme evenings, etc.)

Example of our needs: at the Château de Moulerens - 06 to 11 May 2019

- 35 children and young adults, from 2 months to 29 years
- 150 people
- 40 volunteers
- 850 meals
- 60 rooms
- 15 consultation rooms + activities
- 3 conference-exchanges
- 76 individual consultations
- 5 discussion groups

The 2019 gathering at Château de Moulerens (Bordeaux)

Conferences - exchanges and individual consultations

Just an overview of the conferences and individual consultations, each child has its colour! Impressive, isn't it?

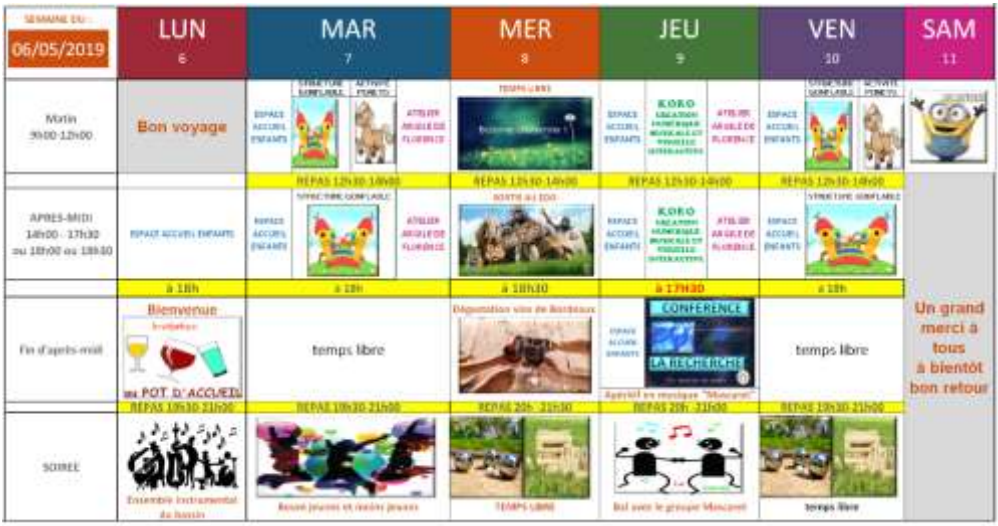
And the schedule was kept, leaving the necessary time for each family to express themselves to the professionals. It is an intense moment, very emotional for each new family, but also for us, the carers. But in the end, it lifts the batteries of many, of the families, but also of the members of the office and the volunteers. Without the carers, it would be impossible to have a successful gathering. They help us with transport, reception, childcare, activities and lots of other things.

		ACCUEIL		GARDERIE	ACTIVITES				
		réception	installation						
LUNDI									
	après-midi	Nelly, Mimie	Isorée, ?	Chantal, Luc, Christine C, Serge C, Geneviève, Hella					
MARDI	matin			Isorée, Madeleine, Mimie, ?, ?	Michel, Hella, Geneviève	Chantal, Luc	Marie		Catherine ?
	après-midi			Chantal, Luc, Marie, Michel, Christine C	Isorée, Serge C, Mimie		Madeleine		
MERCREDI	matin								
	après-midi								
JEUDI	matin			Luc, Eloise, Sophie, Marie, Michel, Christine C			Chantal	Isorée, Charles	Catherine Severine
	après-midi			Isorée, Charles, Sophie, Michel, Eloise, Mimie			Christine C	Chantal, Luc	
VENDREDI	matin			Chantal, Luc, Isorée, Mimie, Hella, ?	Michel, Sophie, Marie	Charles, Eloise			Catherine ?
	après-midi			Isorée, Eloise, Sophie, Marie, Michel, Mimie	Chantal, Luc, Charles				

For 2023 several tracks, if you want to live it for a day, half a day, an hour, just to come and play or to help during meals! Write to us, the success of such an event owes a lot to the carers. **Many thanks to the carers**.

A wide range of activities organised by Sylvie MANTEL

But a gathering is not only clinical or psychological time, it is a set of moments spent together, between friends, to get closer, to discover common points, to talk to each other, to know each other's experiences. And it is Sylvie who takes care of the research of the activities, the



organisation, the planning. Impressive too!!! don't you think?

The Solidad's association, of which several parents of the association are members, made a journey by bike with the arrival at Moulerens castle for the 2017 gathering. They raised funds, part of which helped us to finance an inflatable structure. A great success with the children!

The 2021 and 222 gatherings via ZOOM, COVID oblige

Our loyal professionals

AP-HM La Timone in Marseille

- **Dr Sabine SIGAUDY**, paediatric geneticist at the AP-HM, expert in reference centres

Bordeaux University Hospital

- **Prof. Didier Lacombe**, paediatric geneticist at the Bordeaux University Hospital, expert on Costello syndrome, director of an Inserm unit
- **Dr Didier GRIFFITHS**, dental surgeon at the genetic department of the Bordeaux University Hospital and in private practice
- **Eva TOUSSAINT**, psychologist at the Bordeaux genetic service
- **Rodrigue Rossignol**, Inserm researcher

Geneva Hospital

- **Dr Armand Bottani**, geneticist at the Geneva Hospital, recently retired.

Montpellier University Hospital

- **Pr Didier BESSIS**, dermatologist at the Montpellier University Hospital

Robert Debré Hospital in Paris

- **Pr Alain Verloes**, paediatric geneticist in charge of the genetic department at Robert Debré, has been carrying out Noonan screening since 2002, then on the specificity of RASopathies and leukaemia; coordinates the whole European network of developmental anomalies, which leads to the updating of the PNDS.
- **Dr Yline CAPRI**, paediatric geneticist at Robert Debré
- **Faustine AGEORGES**, psychologist in the genetic department of Robert Debré

Toulouse University Hospital

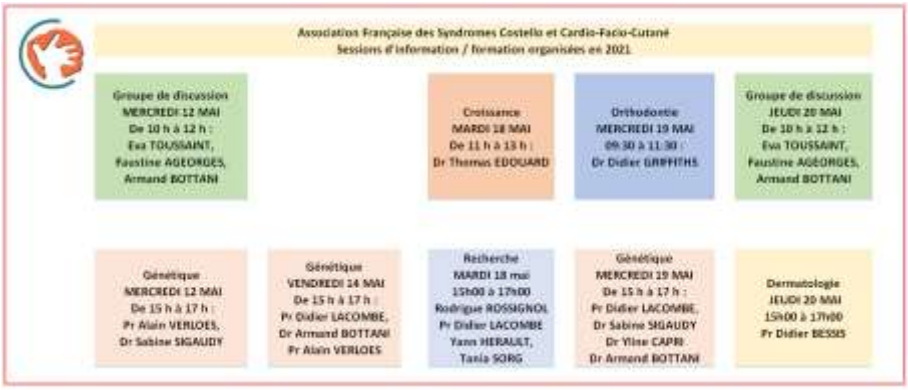
- **Pr Thomas EDOUARD**, paediatrician at the Children's Hospital of the Toulouse University Hospital, member of the endocrinology, bone diseases and genetics department, research project on Noonan syndrome.

Institute of the Mouse Clinic in Strasbourg

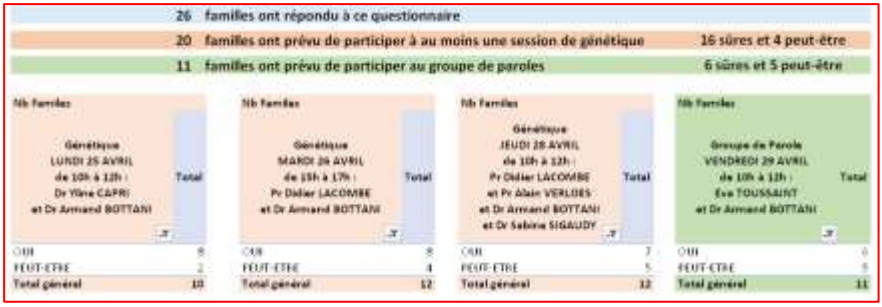
- Tania SORG, Chief Operating Officer
- Yann HERAULT, Director of the ICS

Answers and always the joy of seeing each other again, even on video

2021



2022



A booklet of answers produced by the families, in several languages

Here are the topics covered and an overview of the issues related to the syndromes.

Do not hesitate to ask for it, by email or post. 26 pages, there is a lot of material!

- **Syndromes: cause, heredity, diagnosis**
- **Follow-up, becoming an adult**
- **Epilepsy, behavioural disorders**
- **Dermatology**
- **Growth, puberty, bone and muscle in Rasopathies**
- **Other topics covered**
- **Other testimonials**
- **Informing oneself, informing professionals**

Thanks to the families and to Lesley and Michel PERRIN who provided the translations.

Four *excerpts from the more than 120 questions and answers*

2. The syndromes : causes, heredity, diagnosis 7

2.1. Cause of CFC syndrome : is it hereditary ? 7
2.2. What causes gene mutations ? Is there an environmental cause ? 7
2.3. In genetic terms, making a child is a very complicated process, hence possible accidents 7
2.4. Are non-interpretable mutations communicated to parents ? 7
2.5. Are north african populations really more at risk of incurring genetic mutations or diseases ? 7
2.6. From what moment of pregnancy is a screening test offered ? 8
2.6.1. The parents already have a child with Costello or CFC 8
2.6.2. In the population at large 8
2.7. Can we detect these diseases during pregnancy by observing the behaviour of the foetus, or something specific ? 8
2.8. Is the diagnosis of particular gravity accepted when there is a strong warning sign of early prenatal cardiomyopathy? 8
2.9. Is screening for Costello syndrome or CFC possible and recommended in parents' siblings? 9
2.10. Case of a baby with Costello syndrome diagnosed antenatally in Germany. Will such diagnoses become

3. Follow-up, becoming an adult 12

3.1. Who can take charge of the follow-up? with what regularity ? 12
3.2. What follow-up is required for CFC ? 12
3.3. Adult follow-up in Paris region 12

5. Orthodontics 14

5.1. One aspect of the difficulties of orality 14
5.2. To be able to "treat", it is necessary to work on awareness on a daily basis and to ensure familiarity 14
5.3. A hygiene problem for a weakened mouth. How do you go about it? 14
5.3.1. Plaque ? Role of toothpaste. Setting up a ritual 14
5.3.2. Be vigilant about eating habits, compensate for acids 14
5.4. Care 14

6. Dermatology 16

6.1. Skin problems 16
6.2. Testimony of a 22 years old adult 16
6.3. Why is it so demanding ? 16
6.4. Are dermatological problems due to psychological problems ? 16
6.5. Tendency to haematomas ? Swollen knuckles ? 16

Souvenir photos from the 2019 gathering

All together! What beautiful complicities…

Louise and Lisa

Malie carrying Milena

Stella and Niel

Suzon, volunteer and Louise

Jonathan, Stella, Lisa and Malie

Elisa and Malie

➔

One evening, the children invite us to their show!

The show is created by a teamwork helped by Izorée...

And ends with a nice curtsy...

They were all very studious during a conference-exchange.

But they also know how to rest, or play bowls!

And dancing on the night of the boom, isn't that Lisa and Malie!

Or like Benoit, stay calm.

And always have fun with your brothers and sisters...

Good times with friends who get together

David and his wife, what memories to share with Sylvie.

So much for Anne-Marie and François

But do you recognise them? No ? Our faithful professionals who share meals with us!

Two of the three families who created the association together, Séverine and Théo, and Christine. The only thing missing was Carmen! We'll be back in 2023 !!! Laure impassive while listening to her favourite music.

At Moulerens, we always offer pony rides, a very soothing time, appreciated by all, carriers of a syndrome but also for brothers and sisters.

With the GULLAMBEAU pony club of Gradignan

And this year there was a face painting activity!






Meals are a great moment of sharing, to create links and to welcome new families.

They support us

OPEN GARDENS, visiting gardens for the benefit of associations

For several years, all over France, members of the OPEN GARDENS association have been preparing their gardens all year round to open them to the public. You can visit them,

OPEN GARDENS / JARDINS OUVERTS
Jardinage et Esprit de Générosité

enjoy the peace and quiet, buy some plans, taste homemade cakes, and thus support associations.

Each year, thanks to Lesley and Michel Perrin from PORTETS who support our association, Mr Mick MOAT, president of OPEN GARDENS, gives us a significant sum to help us finance the next meeting.

In 2023, rendez-vous **aux jardins ouverts from 2 to 4 June 2023** throughout France.

So take this opportunity to meet them, make yourself known and thank them on our behalf!

The volunteer runners, Cédric VANHEE and GSK

Gaspard

Our son Gaspard was born in Lille on 08 August 2014. Since his birth, he had to be followed in intensive care for severe cardiac disorders (chaotic tachycardia), and feeding problems. Some of his physical characteristics (hands, ears ...) directed the geneticists' research towards RASopathies. The diagnosis was announced in early October: Costello G12A syndrome.

Unfortunately, this announcement did not lead to a solution to improve Gaspard's health, as his symptoms were too severe.

On 14 October 2014, Gaspard unfortunately passed away, surrounded by his entire family. His struggle was overwhelming for us, his strength, his fighting spirit, his courage made him our daily superhero: hence his nickname of SuperGaspard!

It had been more than 10 years since I had done any sport, and I felt the need to put on my trainers again...

GSK and the Solidarity runners

I work for a big pharmaceutical company, which has a programme called Coureurs Solidaires, run by the GSK Foundation.

The latter gave me the chance, from October 2017 onwards, to run for the Costello association, and thus fund donations according to the km covered in official races.

This system motivated me to enter longer and harder races, and I went from 10km road races to marathons or trails up to 65km.

From being a solidarity runner, but a solitary one with my little poster pinned to my chest, I was lucky enough to drag some motivated colleagues along with me and T-shirts were made by the GSK foundation.

We are now a team of **15 runners and 2 walkers**, and **we have covered 821 km in 2018**: Jean, Adrien, Emilie, Denis, Nicolas, David, Younes, Benjamin, Stephanie, Virginie (both!), Delphine, Marine, Marie Josée, and Jeremy.

The GSK Foundation was unfortunately dissolved in 2018 and refocused its sponsorship activities in other forms.

In January 2022, after almost 2 years of COVID, the race organisers are slowly starting to get back to work. This is precisely the moment I chose to propose to our new director, who is a passionate runner, to set up a team of Solidarity Runners. I had previously canvassed about thirty people, who were in the starting blocks to resume running... The project was immediately accepted! Naturally, I propose the French Association of Costello & CFC Syndromes as beneficiary, and let's also choose Les Délices de Léa which is dear to my colleague Francis.

The Coureurs Solidaires project is off to a flying start and is fully integrated within GSK, including a Solid'air. Together we raised over 1600 euros.

After 8 months, the Coureurs Solidaires team is: **56 runners or walkers, 87 races completed, 183 race numbers, 1 event on site gathering more than 400 participants, 5000 km, 5000 euros.**

Cédric VANHEE

On 17 and 18 November, we were invited to the GSK production site in Saint-Amand-Les-Eaux to receive the GSK donation in the presence of

Maria Rigotti, Vice President of Quality at GSK Vaccines and Scott McColm, Vice President Secondary Operations at GSK Vaccines

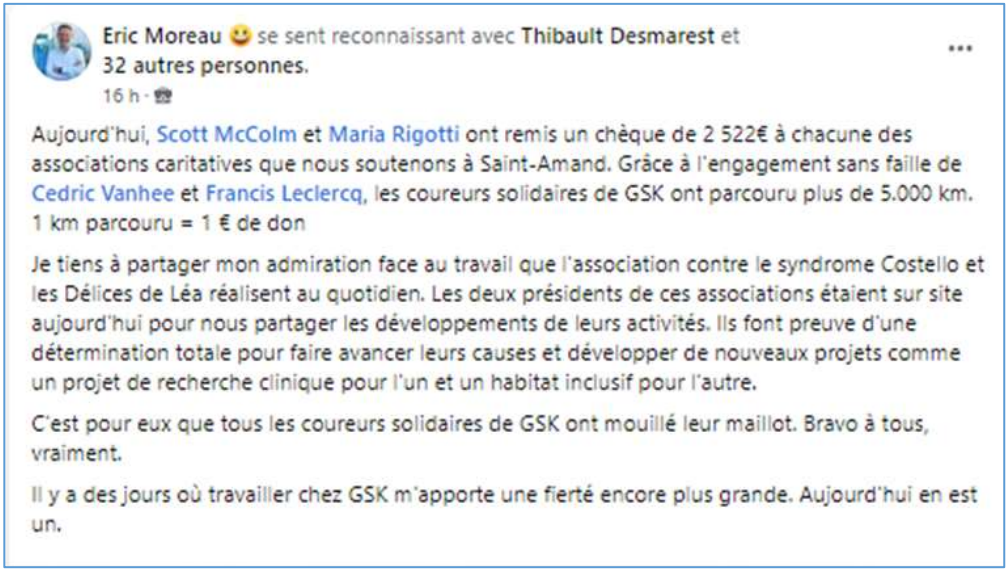
We thank GSK for their welcome, our exchanges and the financial support given to the association.

Testimonies of Cédric's runners and friends

During this donation ceremony, we received a very warm welcome, which makes us want to run with them. Are you ready for 2023?

Contact us!!!

Eric Moreau: feels grateful



Eric Moreau se sent reconnaissant avec Thibault Desmarest et 32 autres personnes.

16 h ·

Aujourd'hui, Scott McColm et Maria Rigotti ont remis un chèque de 2 522€ à chacune des associations caritatives que nous soutenons à Saint-Amand. Grâce à l'engagement sans faille de Cedric Vanhee et Francis Leclercq, les coureurs solidaires de GSK ont parcouru plus de 5.000 km. 1 km parcouru = 1 € de don

Je tiens à partager mon admiration face au travail que l'association contre le syndrome Costello et les Délices de Léa réalisent au quotidien. Les deux présidents de ces associations étaient sur site aujourd'hui pour nous partager les développements de leurs activités. Ils font preuve d'une détermination totale pour faire avancer leurs causes et développer de nouveaux projets comme un projet de recherche clinique pour l'un et un habitat inclusif pour l'autre.

C'est pour eux que tous les coureurs solidaires de GSK ont mouillé leur maillot. Bravo à tous, vraiment.

Il y a des jours où travailler chez GSK m'apporte une fierté encore plus grande. Aujourd'hui en est un.

Christophe Verdier: A lot of energy received during the visit of Serge, the president of the Costello & CFC Association, who was able to give even more reality and concreteness to the kilometres we have travelled to help the association.
A renewed personal motivation in the face of Cedric's enormous motivation for a long time!

Yannick Zagrodnicki: I admire all the work, passion and energy that each member of the association shows every day. Cédric is a true spokesman, and together with Francis, he has relaunched this beautiful initiative of runners in solidarity. As a young runner, and knowing the story of Cédric and Gaspard, I could not but participate in this dynamic...

Well done to all, and I promise,
we challenge ourselves to do even better next year 😊 "
See you soon for a new race

Claire Dusart: "During the Seninghem trail, everyone, caught up in the momentum, went for the longest course...".

What a pride to be part of this Group of Solidarity Runners: We take care of ourselves by doing sport together and as we know that every kilometre covered will give a little more smile to the children who deserve it so much, so we surpass ourselves, we go beyond our limits for them, we let ourselves be guided by those who have a higher level than ours, we stick together and we don't let go, and I can tell you that the shivers we had at the finish line

are indescribable, so deep that we let ourselves be tempted by new challenges that we would never have imagined setting ourselves before. Long live us, but above all long live THEM!

Some souvenir photos

One of the events is the national trail of the Opal Coast.

Do you recognise

Camille and her mum Anne-Sophie who came to support Cédric for OrangeDay?

Virginie Dayez : Small streams make big rivers!

I think we are all proud to be doing our bit and that GSK is committed to us!

Personally it gave me a goal to go beyond my limits and increase distances for a good cause.

Cédric was able to gather his colleagues-friends and he gave it his all!

Depending on the event, not all participants are runners, but walkers who bring their km like Camille and Anne-Sophie!

Contact us to come and run, walk, with them and with us in 2023!

Lyre & Elles

This bouquet of colours invited us to come and listen to this vocal ensemble from Toulouse and its varied and light repertoire.

These six choristers offered us a real bouquet of music from the four corners of the world, from Nougaro to Trenet, from Gospel to Folksong, from Popular Song to Classical Melodies...

They made us love the songs they love and communicated their joy of singing.

Thank you "Lyres et Elles" for having offered us this good musical moment for the benefit of our association.

As you said in your presentation*: "**Music is a wonderful language where all people can meet beyond their differences and divergences**".*

Anne Marie Dupuy

The big Trad ball with MASCARET and LOU REBALEYT in Gradignan

Many of you know the **Mascaret, a** traditional music and dance group from Gradignan.

They came twice to liven up our meetings in Moulerens by making us dance in the meadow.

Stella, like many, appreciates them and accompanies them!

On 3 December, as every year, they are organising a trad dance for the telethon and the Costello-CFC and VHL associations.

Rondeau bordelais, mazurkas, waltzes, bourrées, ... Children, young and old jump together in a good mood, led by about twenty musicians: violins, accordions, flutes, fifes and bohas (pronounced bou-hô) Gascon bagpipes.

This evening is also an opportunity for us to make our association known and perhaps get some new members!

Thanks again to the **Mascaret** for spreading joy around them.

Anne Marie Dupuy

The project for Lea with PACS 1 syndrome

During the presentation by two GSK directors of the donations collected by the solidarity runners, we discovered another association helped by the solidarity runners. Christophe, its president, introduced us to his daughter Léa and her project.

The story of Lea

Léa has PACS 1 syndrome, which has been recognised as a Rare Disease for one year - 12 cases in France and 200 on our planet. With my wife, without medical guidance since her birth, we have helped Léa over the years, so that she progresses and gains in autonomy.

Today I am 25 years old... I am passionate about cooking and I always accompanied my mother in the family kitchen... my playroom! That's how I found my interest.

When I was 20 years old, after leaving the Institut Médico Educatif, I had to find a job... a very big word! As I had no place in an ESAT or other solutions, my parents and I decided to create this professional cooking workshop where we gradually set up learning methods with tools adapted to my difficulties and thus generated the hiring of a person to accompany me, her name is Blandine... I like working with Blandine!

The creation of the associative project "Les Délices de Léa":

Created in 2017, the association project "Les Délices de Léa" aims to include young people with disabilities through a cooking workshop that we have set up in our home

This association includes more than 160 volunteers, who work alongside us throughout the year to reinforce the work in the workshops or to accompany us at external stands.

Our commitment does not stop there, "Trail Evasion en Pays de Lumbres", one of our events, organised with the support of the Municipality of Seninghem and the Communauté de Commune du Pays de Lumbres, more than 2,500 people take part each year to "Run WITH and FOR Extraordinary Children", without expecting any reward, 100% of the profits are donated to associations helping people with disabilities (more than 120,000€ donated since the beginning of the race). A rural village of 750 inhabitants and its Community of Communes are mobilized and committed to our cause! Our reflection :

The initial objective: a professional occupation

The objective was to find for Léa "a professional occupation", so that she could continue to progress (and not regress) like any person in our society, to be useful and recognised. Indeed, "working is not only producing, it is also living together". In this project, beautiful encounters are taking place with other associations and organisations (ELSAA Arques, RPI, Chefs en Nord, 1.2.3. Soleil, Rubans en Pays de Lumbres, les disciples d'Escoffier, etc.), the workshop's clients, stand activities, discovery days with other structures and other young disabled people. The social links are numerous. The diversity of his learning is very wide and continuous. We have seen remarkable progress in the space of a few years and we have also noticed the interest that Léa has in her new life: the joy of getting up in the morning to go to work!

More info at: https://www.facebook.com/lesdelicesdelea1

Want to join us? Please contact us!

The members of the bureau 80

You want to help the association? Please contact us!

Composition of the Bureau elected on 5 February 2022

President Serge ARNOULET

Representative to the Scientific Committee François DUPUY

Secretary Cristina AQUABA Deputy Corinne LEBORGNE

TreasurerNelly PIRIOU Deputy Sylvie MANTEL

Delegates :

- Institutional partners Serge ARNOULET
- Costello syndrome Anahita AVALOS
- CFC syndrome Maïté EDIN
- Inclusion Sergio AVALOS
- Events Georges BAILLY
- Gatherings Sylvie MANTEL

The following were elected to the Young Adults group

to listen to them and to carry out targeted actions

Lisa AOURANE, Lisa BAILLY, Jonathan MAILLARD

If you wish to support us :

By **cheque made out to** A.F.S. Costello & CFC. to **Nelly PIRIOU**

30 Rue des Fossés 03500 SAINT POURCAIN SUR SIOULE,

By a **bank transfer** followed by an email with your details to **contact@afs-costello-cfc.asso.fr.**

IBAN	FR76 1680 6008 2066 0596 2787 381
BIC	AGRIFRPP868

Via Hello Asso, for **a gathering**, HelloAsso-Gathering

For foreign countries, you can use **PayPal** via the "Make a donation" button on our website **http://afs-costello-cfc.asso.fr/**

A **tax receipt** will be sent to you, and if you are taxable, the tax reduction is 66% within the limit of 20% of taxable income.

See you soon and get your encils ready

contact@afs-costello-cfc.asso.fr

French Association of Costello & Cardio-Facio-Cutaneous Syndromes

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