HSP Patients Leader Group
Madrid
2015 June 1

Philippe HANRIAT President	Jean BENARD Vice President Scientific Adviser

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4 GOALS

23 years old

600 HSP affected families

❖❖❖

- **Promote the recognition of our disease** in the French Society especially putting the stress on accessibility
- **Improve our quality of living**
- **Support research** : each year we donate about 25 000 E for HSP research project dealing either with genetics or with physiopathology)
- **Promote the recognition of all rare diseases**, in all country and worldwide

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MEANS: CREATION OF NETWORKS

- **Development : National ASL networking covering districts and France**
- **Integration : French Rare Diseases Alliance Federation**

- **Creation : Federation of Neurodegenerescence Diseases (FND): HSP, French Associations Friedreich & Cerebellar Syndrom**
  - **Common Scientific Committee**
  - **Common Medical & ParaMedical Committees**

  recommendations and guidelines
  - **2 supports for quality of living :**

  ☎ **Psychological phone line run by 2 psychologists entirely devoted to neurological syndromes**

  ☎**Phone line of management for social administrative issues.**

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COMMUNICATION & EDITION

- **Web Site** (forthcoming reappraisal)
- **Magazine "*Spastic*", twice a year (forthcoming is N° 74)**
- **News Letter every 2 months**
- **Booklet "*To live with HSP*" devoted to all aspects of HSP , in press to be issued oct 2015, sponsored by GrOUPAMA insurances Fundation**
- **Annual GA meetings in Lyon, France**
- **Common meetings with FND on Ethics & Quality of Living**

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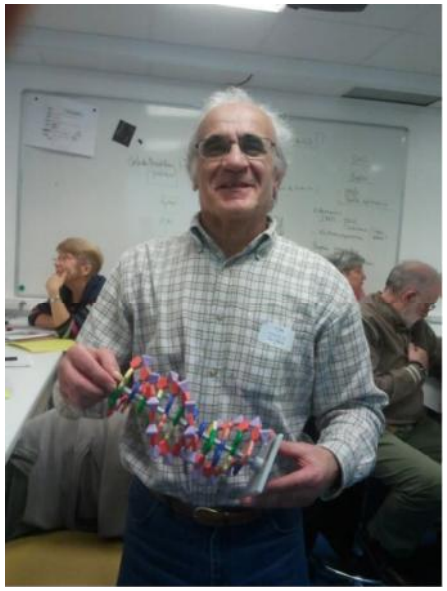
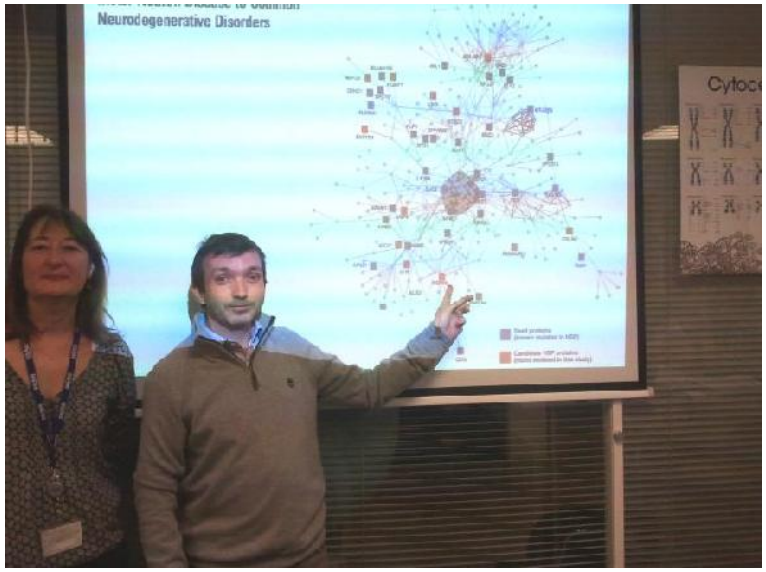
The DNA School at GENETHON



Double helix contemplation



Pr Giovanni STEVANIN at work…

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The ASL DONATIONS to RESEARCH TEAMS 2007 > 2015

YEAR	RESEARCH TEAM	PROJECT	DONATION (€)	RESULT or CONTRIBUTION
2007	Dr Amir BOUKRIS (Pr Giovanni STEVANIN lab, Salepetrière) Paris	Recessive *SPG* Genes identifications	25 000	***SPG* 11 & *SPG*15 identification**
2008	ASL Research Prizes Dr Coralie FASSIER & Anne TARADE (Pr Judith MELKI lab) Evry		4 000	Establishment of a *SPG4 -/-* mice strain C.Fassier *et al.* **Microtubules-targetting drugs rescue axonal swellings in cortical neurons from Spastin-knock-out mice** *Dis. Model. Mech.* 2013
2008	Laboratory of Dr Jamilé HAZAN, Pierre & Curie University Paris	Instalation of the lab: apparatus and reagents	10 000	
2009	Laboratory of Dr Jamilé HAZAN, Pierre & Curie University	Instalation of the lab: apparatus and reagents	25 000	
2010	Laboratory of Dr Jamilé HAZAN, Pierre & Curie University	Partial salary of Dr Coralie FASSIER post-doc: *SPG3A* physiopathology	25 000	***SPG3A* physiopathological mechanism: BMP signalling pathway at work**. *Nature NeuroScience* 2010
2011	ASL Research Prize to Drs Jamile HAZAN & Coralie FASSIER	From genes to physiopathological mechanisms	8 000	**Outstanding contributions: Discoveries of *SPG4* and sorting out physiopathology mechanisms associated to SPG3A & *SPG4***
2011	Laboratory of Pr Cyril GOIZET , Bordeaux	Energetic, mitochondrial, and lipidic metabolism in PSH	17 000	
2012	Laboratory of Pr Cyril GOIZET , Bordeaux	Energetic, mitochondrial, and lipidic metabolism in PSH	17 000	First step project towards the *SPG5* gene French project
2014	Laboratory of Dr Jamilé HAZAN, Pierre & Curie University	Disposables & Reagents for *SPG4* physiopathology project	8 000	Allison R., **An ESCRT–spastin interaction promotes fission of promotes fission of recycling tubules from the endosome** *J Cell Biol* . 2013
2014	Laboratory of Dr Fany MOCHEL & Pr Alexandra DÜRR ICM Salepetriere Paris	Metabolic therapeutic intervention for *SPG5* HSP patients	20 000	
2015	Laboratory of Dr Fany MOCHEL & Pr Alexandra DÜRR ICM Salepetriere Paris	Metabolic therapeutic intervention for *SPG5* HSP patients	20 000	
			179 000	

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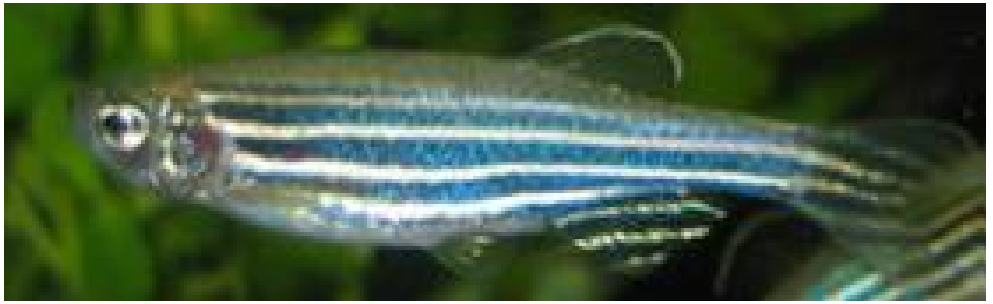
The FORTHCOMING DONATIONS to FRENCH RESEARCH TEAMS 2016-2017

2016	Dr Coralie FASSIER (Dr HAZAN lab) on going request Not evaluated yet	HSP related to *SPG4* locus :physiopatholy and potential therapeutic targets	**20 000**
2016	Dr Guillaume BANNEAU (Pr Giovanni STEVANIN lab) on going request not yet approved by Administrative Committee	Partial Genotype/Phenotype Interface for European countries belonging to SPATAX Network	**6 000**
2017	Dr Coralie FASSIER (Dr HAZAN lab) on going request Not evaluated yet	HSP related to *SPG4* locus :physiopatholy and potential therapeutic targets	**20 000**



Zebra Fish

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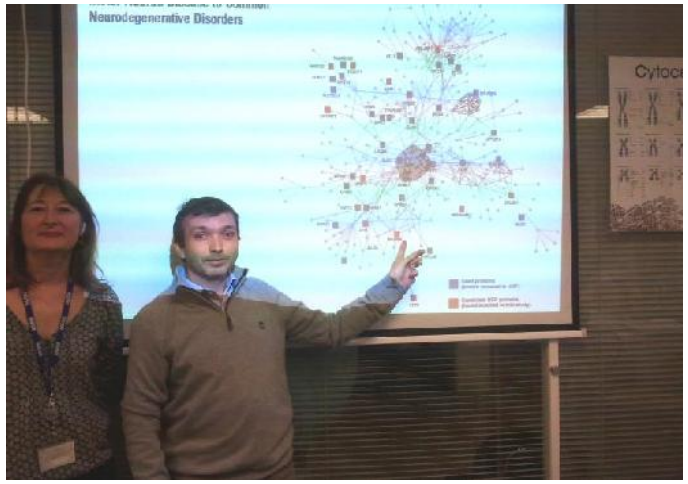
Our RESEARCH TEAMS

SPATAX meeting , Paris 2012

Dr Coralie FASSIER & Dr Jamilé HAZAN
Paris
SPG3A, SPG4 Physiopathology

Pr Cyril GOIZET,
Bordeaux
Energetic
Metabolism



Pr Giovanni STEVANIN, Paris
Gene identification SPG11 & SPG15

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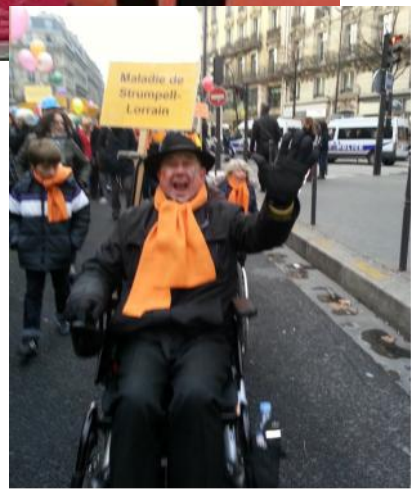
DNA School students

The President
& the Vice President



FND Federation

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CURRENT ASL GOALS & EXPECTATIONS

- ***Use of genetic identification for progeny project of parents***
  - ⇨ **Prenatal testing**
  - ⇨ **In vitro fertilization and embryos testings**
- ***Improvement of HSP Patients care and living***
  - ⇨ **Accessibility**
  - ⇨ **Intercontinental good practices regarding physical training**
- ***Towards targeted treatment for patients***
  - ⇨ **SPG4 deficient**
  - ⇨ **Metabolism deficient**

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Jean BENARD ABILITIES
« PRO and CON »

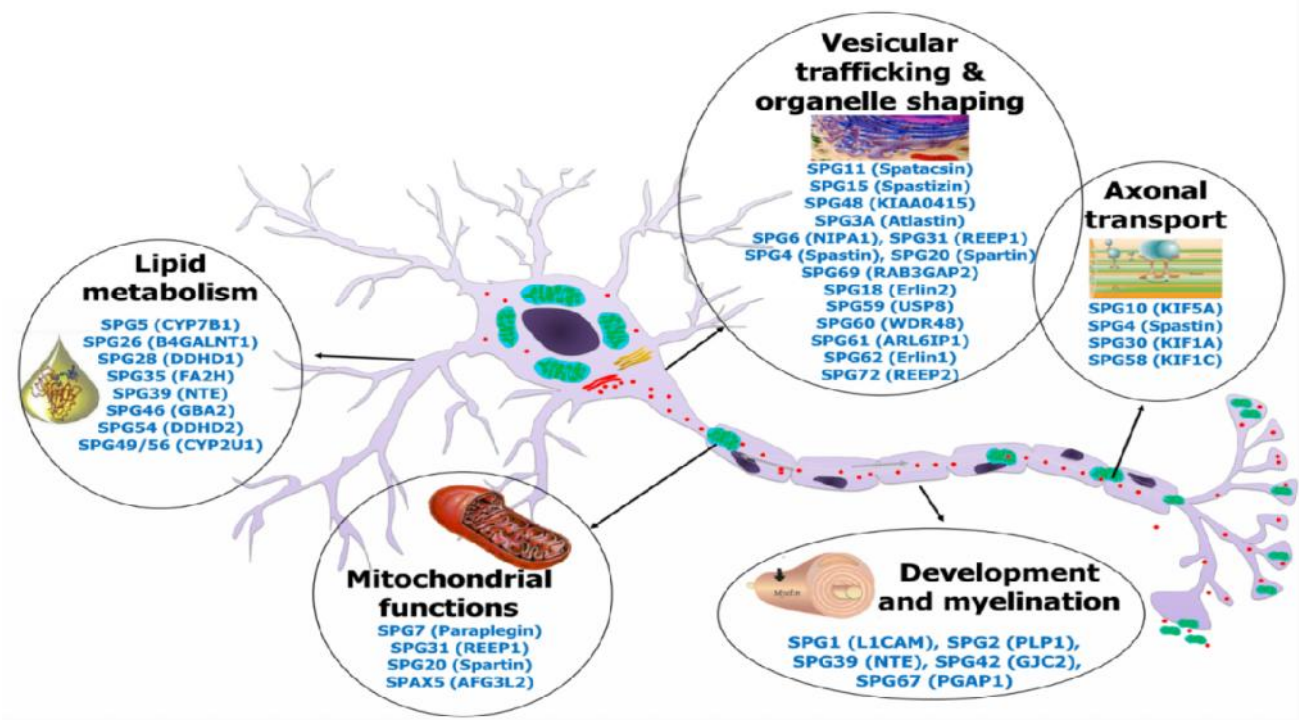
CON

- ***« I am not a business man and I am very bad fund raiser »***

PRO

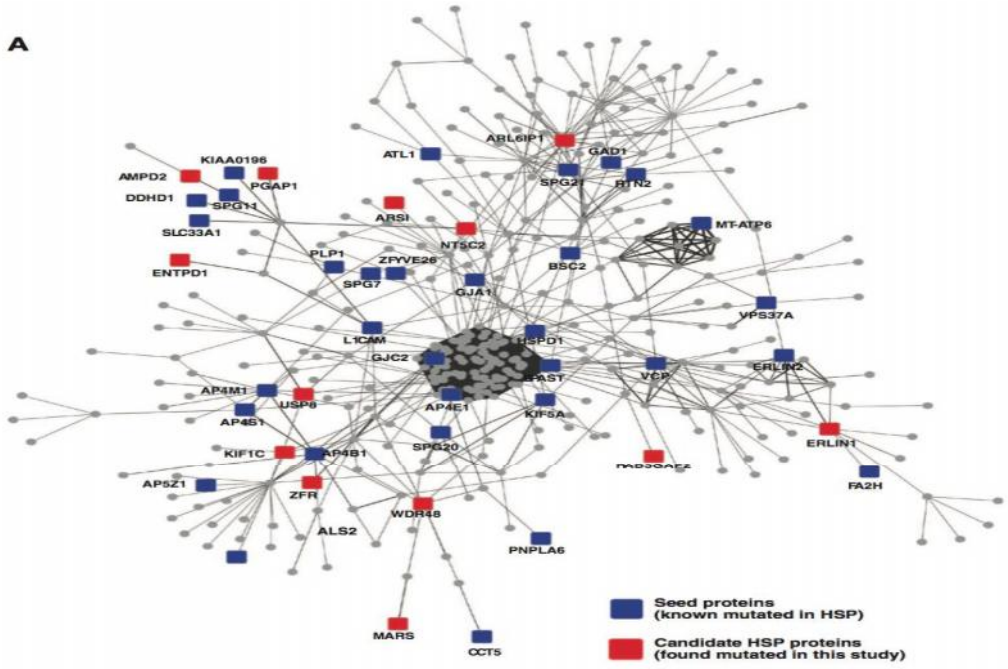
- ***« Eager to afford my own to the HSP battle***
- ***Diplomat: able to put people working together***
- ***Perfectly aware of advances in HSP researches, able to afford advices on strategic approaches »***

➢Extreme genes heterogeneity reflects distinct physio-pathological mechanisms at work in the cortico-spinal neurone



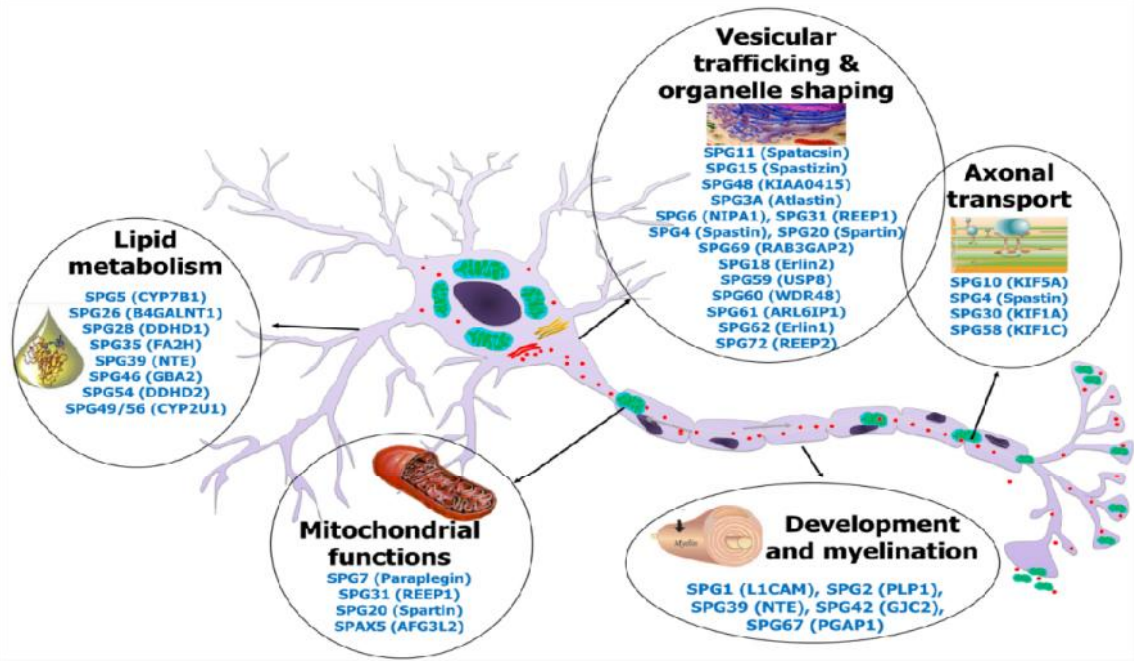
Physiopatology mechanisms from 34 genes

Klebe, Stevanin & Depienne. Rev. Neurol (in press)



Novario G. et al, *Science* 2014

- ***SPG*-encoded proteins, to exert their biological activity, interact between themselves or with other proteins**
- ***SPG*-encoded proteins form a biological network that, if not functional, also operate in other neurological diseases: cerebellum syndrome (CS) or Amyotrophic Lateral Syndrome. A real advantage for fund raising: the more, the best !**
- **This can explain, in families, variations of clinical expression and may reflect observed "passage" from HSP to CS**



➢ **Multiple therapeutic approaches that target the physiopathological mechanism at work, depending on the involved defective *SPG* gene**

➢ **Consequently, distinct cohorts numerous of patients must be obtained to test any therapeutic proposal**