Introduction

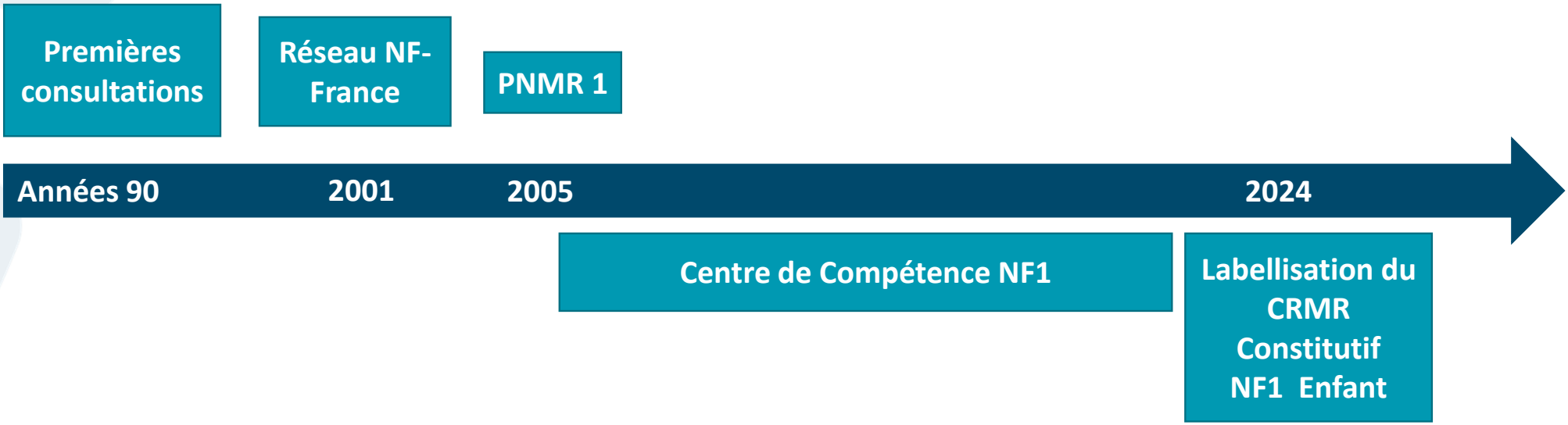
Brève Histoire du Centre

Pr Yves Chaix, Coordonnateur

Journée d'inauguration du CeReNeF'Ped - Vendredi 4 juillet 2025

CHU TOULOUSE

Une *longue aventure* de 30 ans



Histoire du Centre de Référence

Un soutien *indéflectible*

Jean Michel Dubois
(2010-2025)

Anne Henrion
(1998-2010)

La Recherche *dès le début*

MENTAL RETARDATION AND DEVELOPMENTAL DISABILITIES
RESEARCH REVIEWS *2: 48–53 (1996)*

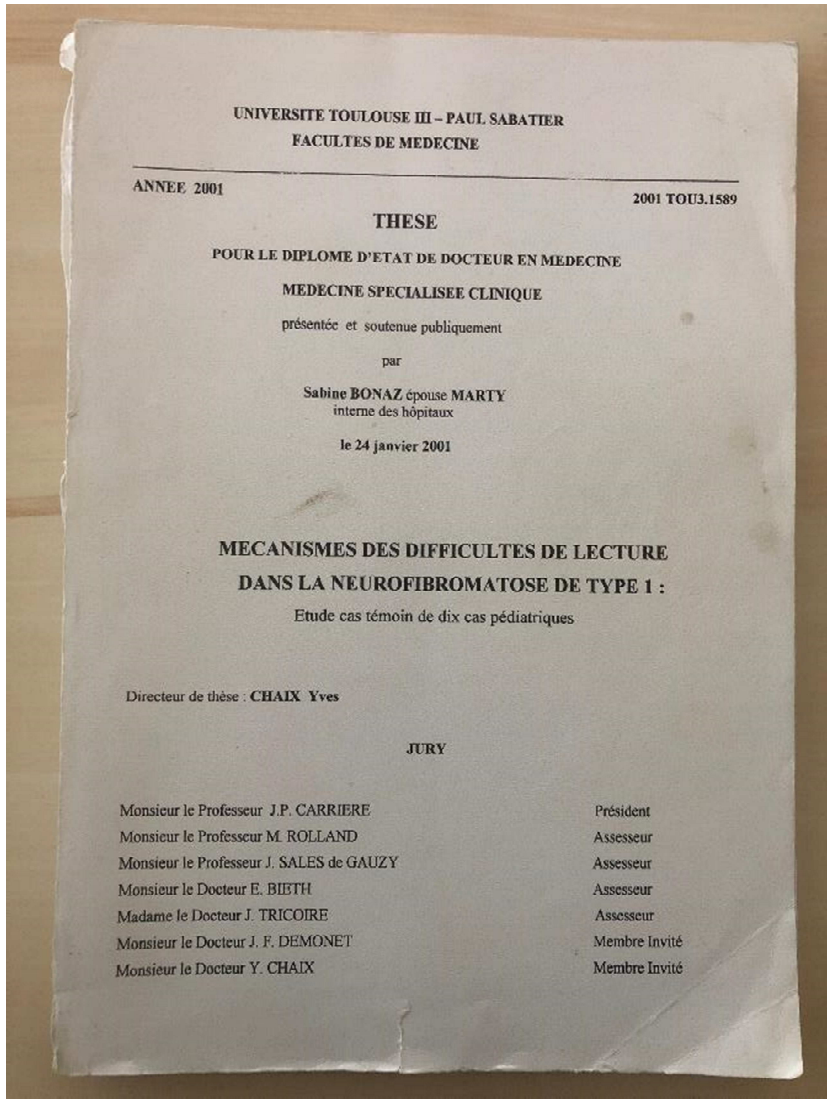
NEUROFIBROMATOSIS TYPE 1: A MODEL FOR THE PATHOGENESIS OF READING DISABILITY

Martha Bridge Denckla
Johns Hopkins University School of Medicine, Kennedy Krieger Institute, Baltimore, Maryland

It is known that neurofibromatosis 1 is associated with lower IQ in affected individuals than in their unaffected relatives. This disorder once was thought to be a model for Rourke's syndrome of nonverbal learning disabilities, but recent research has revealed that the cognitive phenotype is broader, encompassing verbal, linguistic, and reading impairments.

Visuospatial deficit has been confirmed as part of the NF1-associated phenotype, but does not appear to lead to mechanical arithmetic or social failure as Rourke would predict. The reading disability found with NF1 occurs within a developmental language disorder, a context commonly encountered clinically but rarely accounted for by research. Because magnetic resonance imaging yields subcortical rather than cortical findings on children with NF1, this neurogenetic syndrome draws attention to the linguistic contributions of basal ganglia and cerebellum.

one can use any or several of the extant definitions of learning disability as the outcome without reclassifying, for purposes of genetic analysis, who is "affected." Another advantage is that, within the genotype, variation will be visible and measurable at the intermediate level, the imaged brain, thus providing an opportunity to investigate "dose-response" relationships of brain anomalies to cognitive outcomes. Finally, there is a set of purely developmental neurogenetic disorders; that is, their phenotype does not imply deterioration, but follows a curve to what seems to be a stable plateau of delayed or deviant unfolding of brain functions. The study of such disorders permits us to examine critical nature-nurture interactional processes over time.



UNIVERSITE TOULOUSE III – PAUL SABATIER
FACULTES DE MEDECINE

ANNEE 2001 — 2001 TOU3.1589

THESE

POUR LE DIPLOME D'ETAT DE DOCTEUR EN MEDECINE

MEDECINE SPECIALISEE CLINIQUE

présentée et soutenue publiquement

par

Sabine BONAZ épouse MARTY
interne des hôpitaux

le 24 janvier 2001

MECANISMES DES DIFFICULTES DE LECTURE DANS LA NEUROFIBROMATOSE DE TYPE 1 :

Etude cas témoin de dix cas pédiatriques

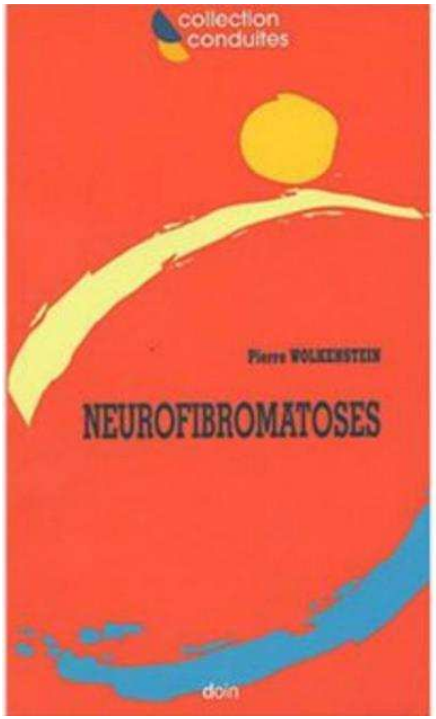
Directeur de thèse : CHAIX Yves

JURY

Monsieur le Professeur J.P. CARRIERE	Président
Monsieur le Professeur M. ROLLAND	Assesseur
Monsieur le Professeur J. SALES de GAUZY	Assesseur
Monsieur le Docteur E. BIETH	Assesseur
Madame le Docteur J. TRICOIRE	Assesseur
Monsieur le Docteur J. F. DEMONET	Membre Invité
Monsieur le Docteur Y. CHAIX	Membre Invité

La Formation *pour tout le monde*

Le Généraliste , 1999



Enseignements :
DES Pédiatrie
Ecoles Psychomotricité
et Orthophonie
DIU Neuropédiatrie
DIU Neurodéveloppement

Chaix Y, Rodriguez D. *Troubles des apprentissages dans la NF1 chez l'enfant.*
In: P. Wolkenstein (Ed.). La Neurofibromatose de Type 1.Doin. 2003, p68-88.

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La Clinique au *centre du dispositif*

NF1
1/3500
Autosomique Dominante
Gène ch 17q11.2
Pénétrance complète
Expressivité variable

Table 1 Comparison of the NIH diagnostic clinical criteria for NF1 from 1988 with the newly revised NF1 diagnostic criteria according to Legius et al. (2021)

Revised diagnostic criteria for NF1 according to Legius et al. (2021)[a]	NF1 diagnostic criteria according to the National Institutes of Health Consensus Development Conference (1988)[b]
(A) The diagnostic criteria for NF1 are met in an individual who does not have a parent diagnosed with NF1 and if two or more of the following features are present:	An individual meets the diagnostic criteria for NF1 if two or more of the following features are present:
• ≥ 6 café-au-lait macules > 5 mm in greatest diameter in prepubertal individuals and > 15 mm in postpubertal individuals [c] • freckling in the axillary or inguinal region [c]	• ≥ 6 café-au-lait macules > 5 mm in greatest diameter in prepubertal individuals and > 15 mm in postpubertal individuals • axillary or inguinal freckles (Crowe's sign)
• ≥ 2 neurofibromas of any type or one plexiform neurofibroma	• ≥ 2 neurofibromas of any type or one or more plexiform neurofibromas
• optic pathway glioma	• optic glioma
• ≥ 2 Lisch nodules identified by slit-lamp examination or ≥ 2 choroidal anomalies defined as bright patchy nodules imaged by optical coherence tomography (OCT)/ near-infrared reflectance (NIR) imaging	• ≥ 2 Lisch nodules
• a distinctive osseous lesion such as sphenoid wing dysplasia [d], anterolateral bowing of the tibia or pseudarthrosis of a long bone[e]	• a distinctive osseous lesion such as sphenoid wing dysplasia or thinning of the cortex of the long bones (with or without pseudarthrosis)
• a heterozygous pathogenic *NF1* gene variant with a variant allele frequency of 50% in apparently normal tissue such as white blood cells	
(B) A child of a parent who meets the diagnostic criteria specified in A meets the diagnosis of NF1 if one or more of the criteria in A are present[f]	• a first-degree relative NF1 (parent, sib or offspring) diagnosed with NF1 by the above criteria

Kehrer-Sawatzki et al., 2022


Protocole National de Diagnostic et de Soins (PNDS)
Neurofibromatose 1

Texte du PNDS

Centre de référence labellisé NEUROFIBROMATOSES

Août 2021

Centre de référence labellisé NEUROFIBROMATOSES / Août 2021
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