POSTGRADUATE MEDICAL TRAINING

FMH Medical Genetics

at the

Service of Medical Genetics,
Department of Gynecology, Obstetrics and Genetics (DGOG)
Centre Hospitalier Universitaire Vaudois (CHUV)
and
Université de Lausanne (UNIL)

Head of the Service : Prof. Jacques S. Beckmann, Professeur ordinaire de la Faculté de Médecine à Lausanne

Clinical geneticists : Dr med. Marie-Claude Addor, FMH for Medical Genetics; Dr med Florence Fellmann, spécialiste en génétique médicale; Dr med Sébastien Jacquemont, spécialiste en génétique médicale; Dr Christian Monnerat, FMH for Internal Medecine and Oncology

Cytogenetic diagnostic laboratory and molecular cytogenetic laboratory : Anne-Claude Gaide, FAMH; Dr Danielle Martinet, FAMH

Molecular diagnostic laboratory : Dr Francine Thonney, FAMH

Unit of cancer cytogenetics : Prof. Martine Jotterand, PhD FAMH; Dr Dominique Mühlematter, PhD FAMH; Dr Valérie Parlier, PhD FAMH; Dr Sarah Porter, PhD, registered clinical scientist

Number of supervisors:	5
Genetic counsellor :	1
Actual number of trainees:	1

(1) Postgraduate Medical training for the FMH Medical Genetics

(A) Description of the postgraduate training centre

Genetic Medicine in Lausanne was first developed in the late 60's by Professor E. Juillard and Dr A. Catti. Their successor was Professor Graziano Pescia, who, in collaboration with the obstetricians of the Maternity, was the pioneer for chorionic villi sampling analysis. Professor Daniel Schorderet acted as interim head of the Division from November 1998 till September 2002. Since then, the current head is Professor Jacques S. Beckmann and the Division was upgraded to a Service. The whole Service of Medical Genetics consists of approximately 50 individuals with main goals of patient care, research and teaching. Its structure is affiliated both with the University of Lausanne's Medical School and the CHUV. In the latter, its clinical and laboratory diagnostic activities are under the auspices of respectively the Department of Gynaecology, Obstetrics and Genetics (DGOG) and the Department of Laboratory Medicine (DML).

(B) Organisation of the Service of Medical Genetics in Lausanne :

1. **Clinical unit** (genetic counselling and diagnostics in nearly all medical specialities : fetal medicine, prenatal diagnosis, dysmorphology/syndromology, malformations, pediatric diseases, complex diseases of the adult, neurogenetics, nephrogenetics, cardiogenetics, dermatogenetics, oncogenetics).

2. **Medical genetics diagnostic laboratories**
   Conventional cytogenetics (pre- and postnatal), Cancer cytogenetics (hemopathies), FISH, CGH, molecular diagnosis (pre- and postnatal), cell culture.

3. **Research groups (of the CHUV's Service and the Unil's Department of Medical Genetics)**

4. **Registry of congenital anomalies in the canton of Vaud (EUROCAT) :** provides essential epidemiologic information on congenital anomalies in our population and is integrated in the European network of the 43 registries of 20 european countries (EUROCAT : "European surveillance of Congenital Anomalies").

The clinical unit covers referrals from the whole canton of Vaud as well as some referrals from other Cantons.

(C) Specialised consultations within the University Hospital of Lausanne (CHUV) and in collaboration with the experts in the respective clinical fields :

- **In Cardiology:** Cardio-genetic consultation
- **In Dermatology**
- **In Gynecology and Obstetrics (**Fetal Medecine, prenatal diagnosis, infertility clinic**)**
- **In Nephrology :** MRG (maladies rénales génétiques)
- **In Neurology :** predictive testing, neurogenetics
- **In Oncology : (**Dr Ch. Monnerat, FMH**)**
- **In Pediatrics** (Neonatology**,** Unit of child development, Neuro-pediatrics, Pedopsychiatry, Pediatric orthopedia)

Apart from these specialised consultations we have referrals from nearly all medical specialities of the CHUV or from the external medical doctors : ophthalmology, ENT, surgery, endocrinology, psychiatry.

(D) Statistics (see attached file)

(E) Postgraduate training for FMH Medical Genetics

We usually employ trainees for 2 years and encourage them to spend the third year in another institute of medical genetics. Coaching conditions are extremely favourable as there are, not counting the genetic counsellor, actually 5 supervisors for 1 trainee.

Mandatory during the two years of postgraduate training :

- **Active participation to all genetic consultations and clinical staff meetings .** Active participation to weekly staff meeting
- **Rotation of up to 3 months in the cytogenetic diagnostic laboratory**
- **Rotation of up to 3 months in the molecular diagnostic laboratory**
- **Regular postgraduate training seminars in collaboration with the Division of Medical Genetics in Geneva** (once a month)
- **Bimonthly staff meetings on dermatogenetics**
- **Monthly meetings on neurogenetics**
- **Weekly nephrogenetics staff meeting**
- **Pedopsychiatric staff meeting**
- **Prenatal staff meeting (twice a month)**
- **REMPOL oncogenetic staff meeting (5 x per year)**
- **„Réunion Vaud / Genève“** (3-4x /year with regular active participation)
- **„Swiss dysmorphology meeting“** (2-3x/year with regular active participation)
- **Participation at the bi-annual meeting of the Swiss Society of Medical Genetics**
- **Once a year participation to an international congress or postgraduate training course of Medical genetics (financed by the CHUV)**

Facultative :

Pediatrics: „Colloque Formation Pédiatrie“ (1x/week)
„Formation générale du CHUV pour les médecins internes de toutes spécialités“
Weekly research lab meetings in the dept of Medical Genetics (UNIL)
According to the personel interest of the assistant

(F) **Educational materials**

- access to the Service's journal and book collection as well as to the main library of the faculty of Medecine in the main building of the CHUV
- Updated books and databases (POSSUM, LDDB-Winter-Baraitser, OMIM, Geneclinics) in the medical office
- Internet-access with free download possibility for nearly all medical journals at each working place

(G) Evaluation of the trainees

The consultants plan regular evaluations (at least twice a year) of the trainees and support their individual carrier planning. In addition the trainees get at the end of their postgraduate training programme an official FMH certificat evaluation.

(2) Postgraduate Medical training for another FMH speciality

Occasionnaly we also get an assistant for a 6-12 months period, a postgraduate trainee in another FMH specialty (mostly in pediatrics or gynecology).

APPENDIX

Data

Consultation type	*Number 2003*	*Number 2004*	*Number 2005*	*Changes with respect to 2004*
Ambulatory (*prenatal diagnosis)*	***970*** **(546)**	**918** ***(539)***	***956*** **(562)**	**+ 4.1%** **+ 4.2%**
Hospital	***88***	***80***	***95***	***+18.8%***
Total	***1058***	**998**	***1051***	***+ 5.3%***

CONSULTATIONS 2005

	2005	2004
Prenatal diagnosis	562	**539**
Mental retardation – dysmorphism – malformations	**226**	240
Genetic counselling	165	**139**
Oncogenetics	76	**57**
Infertility FC	22	**21**
Unknown	**—**	**2**
Total	1051	998

Eurocat - VAUD 2004 - Cases and prevalence per 10,000 births
Denominator :

Total number of births (live and still) : 7092 *Age of mother >=35* 1542
Total number of live births : 7064 *Age of mother <30* 2839

Anomaly Subgroup	LB (n)	FD (n)	IA (n)	LB+FD+IA (n)	LB+FD+IA (rate)
All Cases	**202**	**1**	**70**	**273**	**384,94**
All Cases (2003)	***179***	***0***	***37***	***216***	***314,46***
Nervous system	6	0	19	25	35,25
Neural Tube Defects	1	0	13	14	19,74
Anencephalus and similar	0	0	6	6	8,46
Encephalocele	1	0	4	5	7,05
Spina Bifida	0	0	3	3	4,23
Hydrocephaly	1	0	0	1	1,41
Microcephaly	1	0	0	1	1,41
Arhinencephaly/holoprosencephaly	0	0	1	1	1,41
Eye	2	0	2	4	5,64
Anophthalmos/microthalmos	0	0	0	0	0,00
Anophthalmos	0	0	0	0	0,00
Microthalmos	0	0	0	0	0,00
Cataract	1	0	0	1	1,41
Ear	2	0	1	3	4,23
	1	0	1	2	2,82
Anotia	1	0	0	1	1,41
Microtia	0	0	1	1	1,41
Congenital heart disease	84	1	13	98	138,18
Anomalies of cardiac chambers and connections	3	0	0	3	4,23
Common arterial truncus	0	0	0	0	0,00
Transposition of great vessels (complete)	0	0	0	0	0,00
Single ventrical	1	0	0	1	1,41
Malformations of cardiac septa	72	0	11	83	117,03
Ventricular septal defect	39	0	9	48	67,68
Atrial septal defect	37	0	1	38	53,58
Atrioventricular septal defect	4	0	3	7	9,87
Tetralogy of Fallot	2	0	0	2	2,82
Malformations of valves	13	1	3	17	23,97
Tricuspid atresia and stenosis	0	1	0	1	1,41
Ebstein's anomaly	0	0	0	0	0,00
Aortic valve atresia/stenosis	2	0	0	2	2,82
Hypoplastic left heart	0	0	2	2	2,82
Malformations of the great arteries and veins	19	0	4	23	32,43
Coarctation of aorta	2	0	0	2	2,82
Cleft lip with or without palate	4	0	1	5	7,05
Cleft palate	5	0	6	11	15,51
Digestive system	13	0	6	19	26,79
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis	1	0	0	1	1,41
Congenital absence, atresia and/or stenosis of the small intestine	4	0	1	5	7,05
Congenital absence, atresia and/or stenosis of the duodenal	3	0	1	4	5,64
Congenital absence, atresia and/or stenosis of other specified parts of small intestine	1	0	0	1	1,41
Ano-rectal atresia and stenosis	0	0	2	2	2,82

Internal urogenital system-ovaries uterus and renal system	40	0	8	48	67,68
Bilateral renal agenesis	0	0	0	0	0,00
Cystic kidney disease	3	0	1	4	5,64
Congenital hydronephrosis	6	0	1	7	9,87
Bladder extrophy	1	0	2	3	4,23
External genital system	23	0	3	26	36,66
Hypospadias	4	0	0	4	5,64
Indeterminate sex	0	0	0	0	0,00
Limb	20	0	5	25	35,25
Limb reduction	1	0	3	4	5,64
Upper limb reduction	1	0	3	4	5,64
Complete absence of upper limb	0	0	1	1	1,41
Absence of upper arm and forearm with hand present	0	0	0	0	0,00
Absence of both forearm and hand	0	0	0	0	0,00
Absence of hand and fingers	1	0	2	3	4,23
Longitudinal reduction defect/shortening of arm	0	0	1	1	1,41
Lower limb reduction	0	0	1	1	1,41
Complete absence of lower limb	0	0	0	0	0,00
Absence of thigh and lower leg with foot present	0	0	0	0	0,00
Absence of both lower leg and foot	0	0	0	0	0,00
Absence of foot and toe	0	0	1	1	1,41
Longitudinal reduction defect/shortening of leg	0	0	0	0	0,00
Polydactyly	4	0	2	6	8,46
Syndactyly	3	0	0	3	4,23
Musculoskeletal and connective tissue	16	0	12	28	39,48
Choanal atresia	0	0	0	0	0,00
Craniosynostosis	2	0	0	2	2,82
Pierre Robin Syndrome	3	0	0	3	4,23
Mandibulofacial dystosis (Treacher-Collins and Franceschetti)	0	0	0	0	0,00
Oculomandibular dysostosis (Hallerman-Streiff	0	0	0	0	0,00
Goldenhar's Syndrome	0	0	0	0	0,00
Chondodystrophies and osteodystrophies	0	0	2	2	2,82
Diaphraghmatic hernia	0	0	1	1	1,41
Omphalocele	1	0	5	6	8,46
Gastroschisis	2	0	0	2	2,82
Prune Belly Syndrome	0	0	1	1	1,41
Chromosomal	3	0	46	49	69,09
Down Syndrome	1	0	25	26	36,66
Patau syndrome (trisomy 13)	0	0	1	1	1,41
Edward syndrome (trisomy 18)	0	0	5	5	7,05
Other trisomies and partial trisomies of autosomes	0	0	4	4	5,64
Monosomies and deletions from the autosomes	0	0	4	4	5,64
Turner's syndrome	1	0	5	6	8,46
Klinefelters syndrome	1	0	2	3	4,23
Anomalies outside normal range	2	0	0	2	2,82
All Cases	**202**	**1**	**70**	**273**	**384,94**

LB - Live Births
FD - Fetal deaths / Still Births from 20 weeks gestation
IA - Induced Abortions following prenatal diagnosis
- Data not available

Laboratory for molecular analyses

Rapports d'analyse

Le **nombre de rapports d'analyse** émis est d'environ 448 (459 en 2004).

Indications	2005			*2004*		
	Nombre rapports	**Pt OFAS >=**	**Contr. Qual.**	***Nombre rapports***	***Pt OFAS***	***Contr. qual.***
At. de Friedreich	5	1250	**3**	70	2004	
SCA1	7	1750	**1**			
SCA2	9	2250	**1**			
SCA3	5	1250	**1**			
SCA6	8	2000				
SCA7	7	1750				
DYT1	6	1500				
Chorée de H.	27	6750	**3**			**3**
DRPLA	14	11500				
Surdité	6	1200		9	3600	
CMT1A	3	2100		10	8500	
Tomaculose *				2	600	
CMTX	2	500		0	0	
Alpha-1-antitrypsine	7	2030		8	2300	**3**
FGFR3	10	3500			0	
FGFR1	2	700			0	
FGFR2	2	700		8	2000	
TWIST	0	0		4	1400	
Fièvre méd.	21	4200		10	5000	
Hémochromatose	38	9500	**3**	50	17000	**3**
Mucoviscidose-CFTR	94	47000	**7**	44	35000	**6**
Stérilité-CFTR	18	9000		84	67000	
Prader Willi, Angelmann	18	4500	**5**	10	3500	**4**
X fragile	52	15080		89	26000	**3**
Steinert	29	8410		12	9600	
SMA	7	3150		10	4500	
Norrie	0	0		2	600	
MTM1	0	0		4	1400	
Y microdél.	6	2100	**3**	2	1000	**7**
UPD	1	0		1	400	
Contrôle contamination maternelle	13	3900		5	1500	
Kennedy	0	0		2	500	
Divers	35	10500		1	250	
Sous-traitance	13	0		27	0	
c.q. séquençage		0	**4**		0	
Total rapports	**465**	0	**31**	**365**	0	**29**
Extractions ADN facturables	579	57900		591	59100	
Total pt OFAS		**>=215970**			**250750**	

Autres prestations :	**2005**	**2004**	**2003**	**2002**
Mise en banque sans analyse	101	90	177	
Extractions ADN non facturables	33	7		400
CGH sur chromosomes	27	33	25	
Echantillons envoyés à des laboratoires externes	**208**	**120**	**157**	**155**

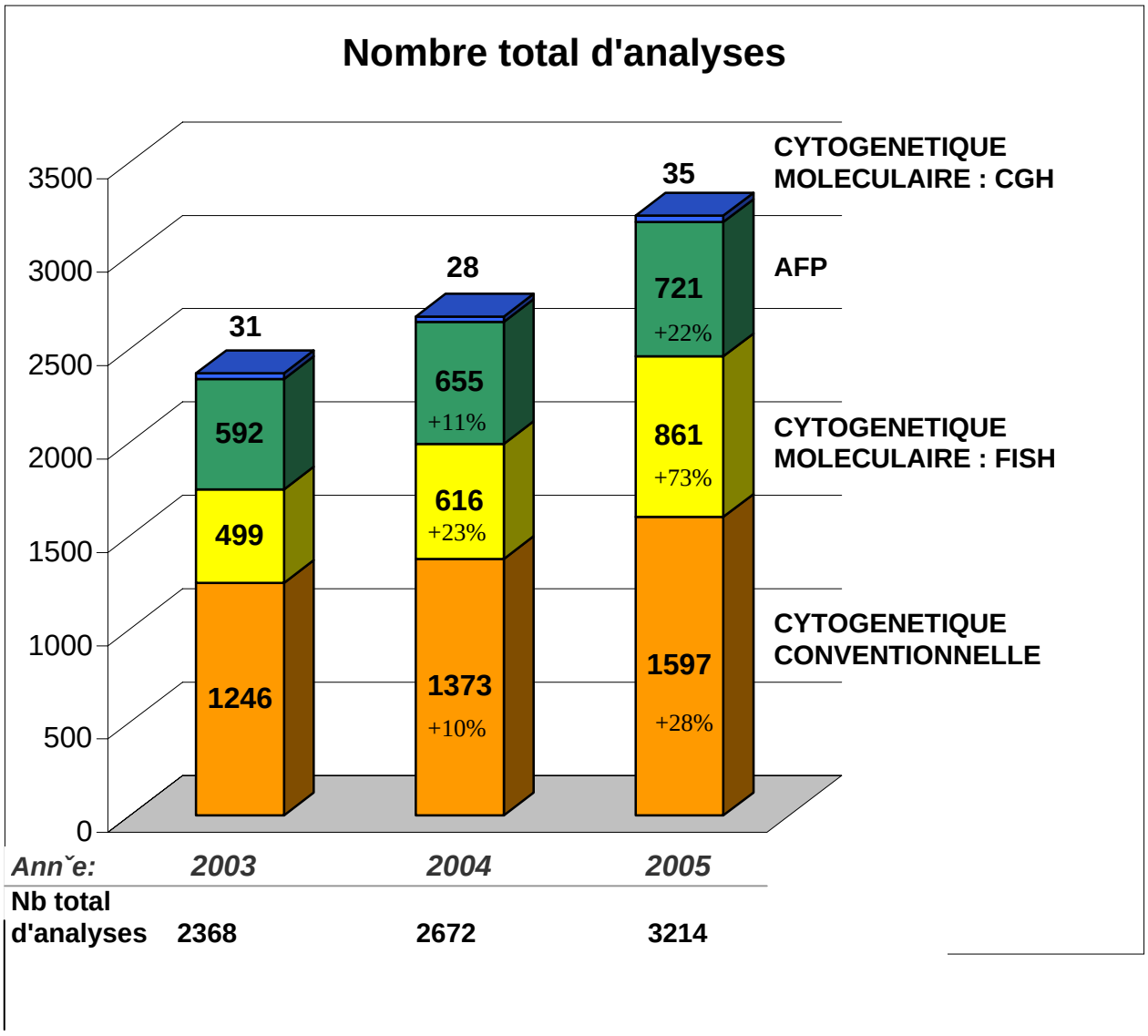
Activité de prestation par technique

Nb Analyses par PCR				**Nb Analyses par Southern**				**Nb Séquençages**	**Électrophorèses capillaires (développement)**
échantillons	séries	échant/ série	contrôles/ série	échantillons	séries	échant/ série	contrôles/ série	séquences	passages
556	191	2.3 (1,2 à 8,1)	2,5	96	16	2.7 (1,5 à 4.3)	2,8	150	38

Laboratory for constitutional cytogenetics

Nbre total d'analyses:

2004	2005	augmentation
2672	3214	**+ 20.3 %**



Evaluation de l'adéquation du nombre d'analyses cytogénétiques et des forces de travail (y.c développement + recherche)

Année	*2003*	*2004*	*2005*	*Augm 2004-2005 (%)*	*Augm. 2003-2005 (%)*
Nbre total d'analyses (cytogén.)	**1776**	**2017**	**2489**	**23**	**40**
Personnel technique	**6**	**5.8**	**7.4**	**27**	**23**
Supervision	**1.3**	**1.5**	**1.5**	**0**	**15**
Nbre total points OFAS	**1'081'000**	**1'229'425**	**1'423'060**	**16**	**32**

Anomalies les plus Fréquemment Observées (Pré- et Post–Natal) :

Les anomalies les plus fréquentes sont en % :

	2004	**2005**
Trisomie 21	**34**	**45**
Réarrangements chromosomiques	**27**	**27**
Trisomie 18	**9**	**13**
Chromosomes sexuels	**16**	**6**

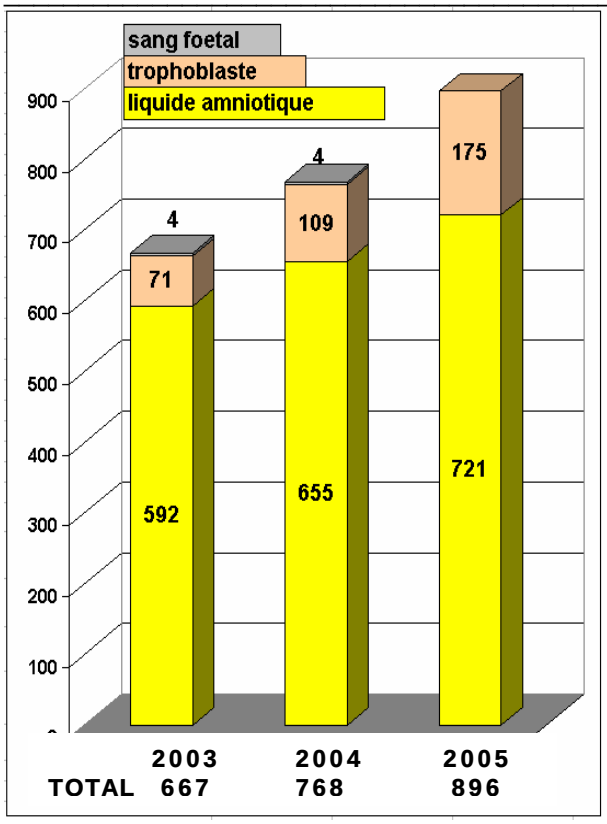
Nombre total d'analyses cytogénétiques :

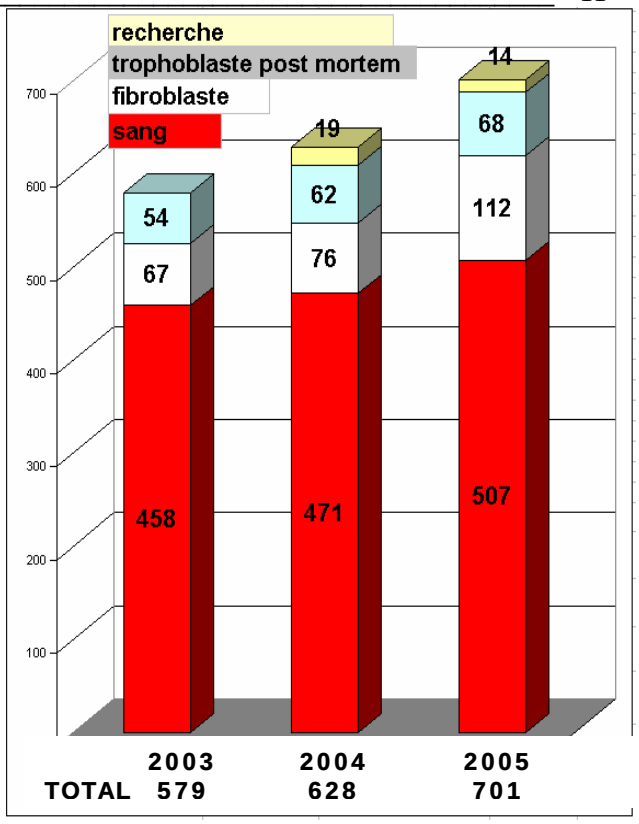
	2004	2005	**augm. %**
analyses prénatales	768	896	**+17%**
analyses postnatales	628	701	**+12%**

analyses prénatales externes	110	217	**+97%**
analyses postnatales externes	190	208	**+9%**

REPARTITION selon les types de tissus

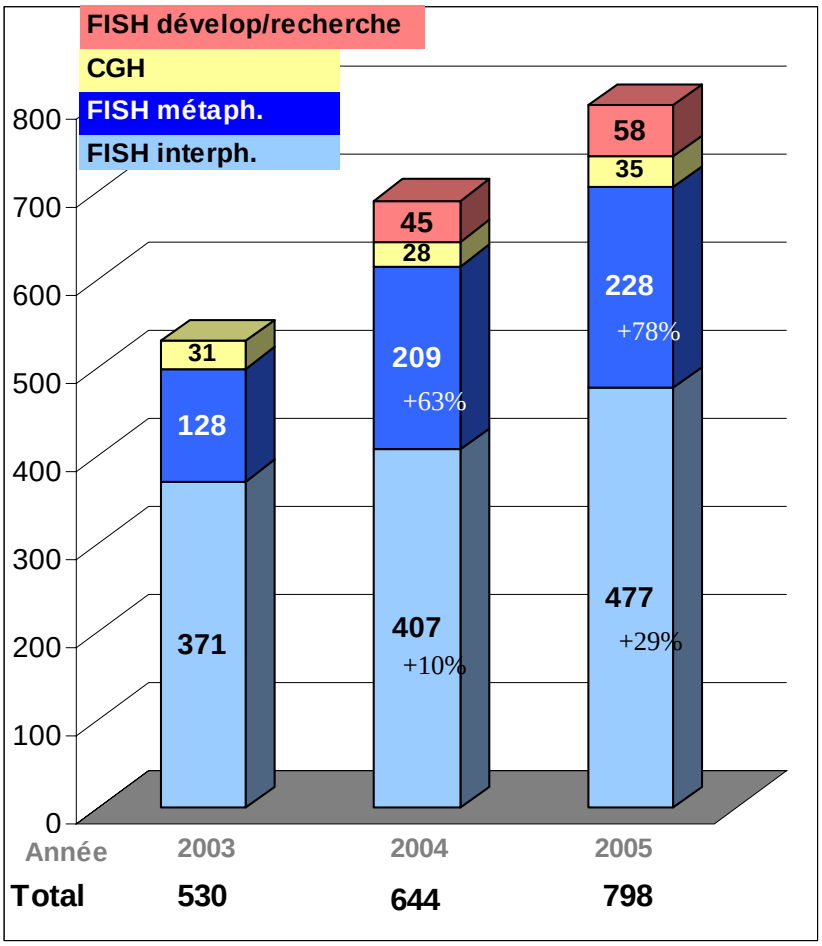
Analyses prénatales Analyses postnatales





Nombre total d'analyses de cytogénétique moléculaire

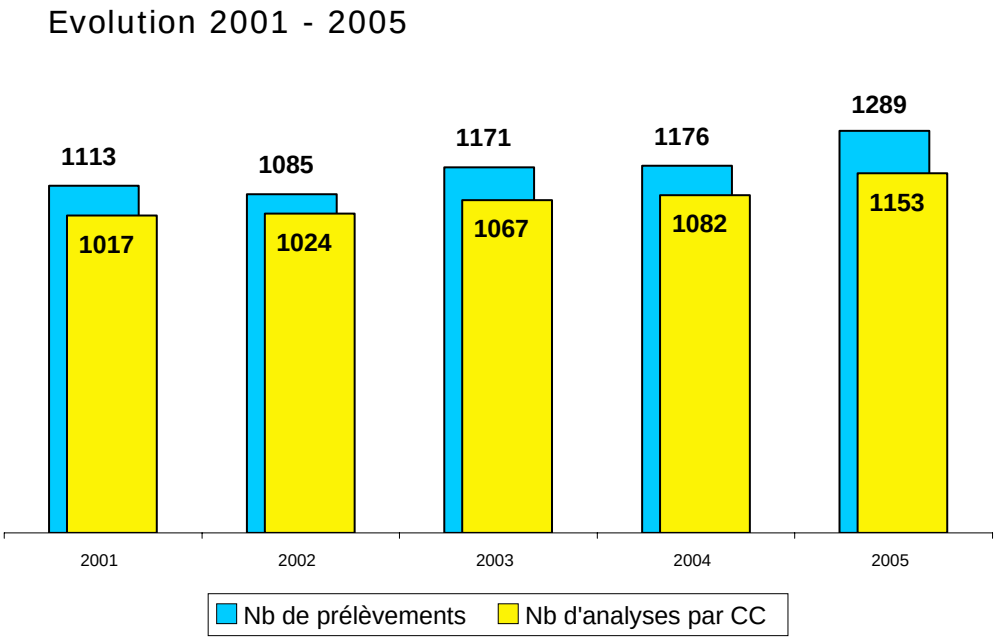
	2004	2005	augm. %
FISH	689	798	**+ 16%**


FISH dévelop/recherche
CGH
FISH métaph.
FISH interph.
800
700
600
500
400
300
200
100
0
31
128
371
45
28
209
+63%
407
+10%
58
35
228
+78%
477
+29%
Année
2003
2004
2005
Total
530
644
798

Cancer cytogenetics unit

Activité diagnostique

Nombre de prélèvements et nombre d'analyses effectuées par cytogénétique conventionnelle (CC)



Analyses effectuées par FISH (avec ou sans cytogénétique conventionnelle) :