

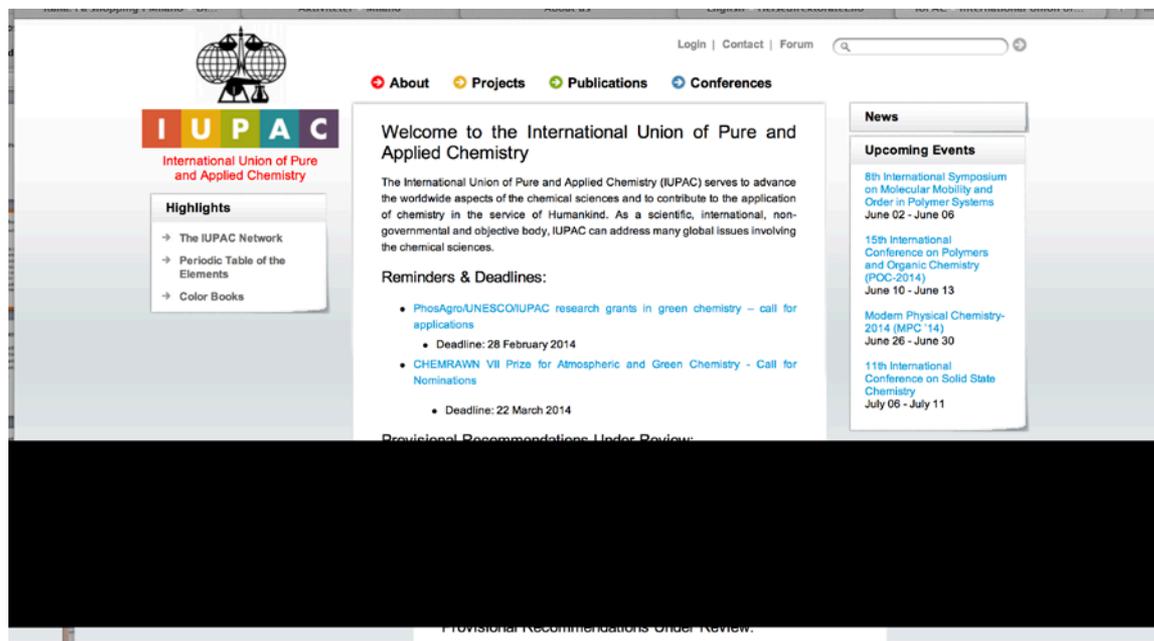
Coding genetic lab tests: A proposal for a logical system

Torunn Fiskerstrand, Consultant, PhD

Center for Medical Genetics og Molecular Medicine, Haukeland University Hospital

National coding system for all laboratory analyses 2004-2014

- Based on NPU - coding system
 - Developed by clinical chemists
 - 16000 codes



The screenshot shows the homepage of the International Union of Pure and Applied Chemistry (IUPAC). The page features a navigation menu with links for 'About', 'Projects', 'Publications', and 'Conferences'. The main content area includes a welcome message, a brief description of IUPAC's mission, and a section for 'Reminders & Deadlines' with two bullet points. A sidebar on the left contains 'Highlights' with links to 'The IUPAC Network', 'Periodic Table of the Elements', and 'Color Books'. A sidebar on the right lists 'Upcoming Events' including the 8th International Symposium on Molecular Mobility and Order in Polymer Systems, the 15th International Conference on Polymers and Organic Chemistry (POC-2014), Modern Physical Chemistry-2014 (MPC '14), and the 11th International Conference on Solid State Chemistry.

IUPAC
International Union of Pure and Applied Chemistry

Highlights

- The IUPAC Network
- Periodic Table of the Elements
- Color Books

Navigation: [About](#) [Projects](#) [Publications](#) [Conferences](#)

Welcome to the International Union of Pure and Applied Chemistry

The International Union of Pure and Applied Chemistry (IUPAC) serves to advance the worldwide aspects of the chemical sciences and to contribute to the application of chemistry in the service of Humankind. As a scientific, international, non-governmental and objective body, IUPAC can address many global issues involving the chemical sciences.

Reminders & Deadlines:

- [PhosAgro/UNESCO/IUPAC research grants in green chemistry – call for applications](#)
 - Deadline: 28 February 2014
- [CHEMRAWN VII Prize for Atmospheric and Green Chemistry - Call for Nominations](#)
 - Deadline: 22 March 2014

Provisional Recommendations Under Review.

News

Upcoming Events

- 8th International Symposium on Molecular Mobility and Order in Polymer Systems
June 02 - June 06
- 15th International Conference on Polymers and Organic Chemistry (POC-2014)
June 10 - June 13
- Modern Physical Chemistry-2014 (MPC '14)
June 26 - June 30
- 11th International Conference on Solid State Chemistry
July 06 - July 11

Laboratory disciplines

- Clinical chemistry
- Microbiology
- Immunology
- Clinical immunology and blood banking
- Clinical pharmacology
- Pathology
- Genetics

Summer 2011

- The existing coding system for genetic analyses
 - Pathcy
 - Without a clear strategy
 - Many codes for the same analysis
 - Inconsistent use of nomenclature and wording
 - One code for each gene
- The result of passive inclusion of all analyses reported back mostly from all laboratory disciplines except medical genetics

Category	ID/code	Component_name	Subcomponent	Method
Molecular bioloy procedure	119	Biobanking_Blood		
Molecular bioloy procedure	81	Biobanking_Cells	Tissue/cell type	
Molecular bioloy procedure	81	Biobanking_Cancercells	Tissue/cell type	
Molecular bioloy procedure	111	Biobanking_DNA	Tissue/cell type	
Molecular bioloy procedure		Biobanking_RNA	Tissue/cell type	
Molecular bioloy procedure	80	Cells_Culture	Tissue/cell type	
Molecular bioloy procedure	43	Extraction-DNA in blood	Tissue/cell type	Method of purification
Molecular bioloy procedure	78	Extraction-DNA in cells	Tissue/cell type	Method of purification
Molecular bioloy procedure	8332	Extraction-RNA in blood	Tissue/cell type	Method of purification
Molecular bioloy procedure	8332	Extraction-RNAnin cells	Tissue/cell type	Method of purification
Category	ID	Komponent_navn	Subkomponent	Metode
Whole genome analysis (screening)		Chromosomal analysis-G banding_Blood	Tissue/cell type	Microscopy
Whole genome analysis (screening)		Chromosomal analysis-G banding_Cells	Tissue/cell type	Microscopy
Whole genome analysis (screening)		Chromosomal analysis-G banding_Tumour cells	Tissue/cell type	Microscopy
Whole genome analysis (screening)		CNV analysis	Tissue/cell type	Platform/array
Whole genome analysis (screening)		Homozygosity analysis	Tissue/cell type	Platform/array
Whole genome analysis (screening)		Gene expression profile	Tissue/cell type	Platform/array
Whole genome analysis (screening)		Genome sequencing	Tissue/cell type	Sequencer
Whole genome analysis (screening)		Exome sequencing	Tissue/cell type	Sequencer
Whole genome analysis (screening)		Methylation analysis	Tissue/cell type	Platform/array/sequencer
Category	ID	Komponent_navn	Subkomponent	Metode
Analysis of spesific chromosomal region		Haplotyping	Locus/Gene name	Platform/array/ microsatellites
Analysis of spesific chromosomal region		Methylation test	Locus/Gene name	MLPA, Southern, sequencing
Analysis of spesific chromosomal region		Chromatin test	Locus/Gene name	ChIP-seq, ChIP-chip
Analysis of spesific chromosomal region		CNV analysis (deletion_duplication)	Locus/Gene name	FISH, MLPA, TaqMan
Analysis of spesific chromosomal region		Chromosomal rearrangement	Locus/Gennavn/type of probe (f.ex breakage, fusion probe etc	
Analysis of spesific chromosomal region		Chromosomal identity	Kromosomnummer	FISH, PCR, FISH-centromerprobe
Category	ID	Komponent_navn	Subkomponent	Metode
Analysis of one gene		Gene analysis - spesific exon(s)	Gene name-number of exon	Sequencing/MLPA
Analysis of one gene		Gene analysis - one gene	Gene name	Sequencing/MLPA
Analysis of one gene		Gene analysis - several genes (gene panel)	Syndrome /disease and Gene names	Sequencing/MLPA
Category	ID	Komponent_navn	Subkomponent	Metode
Analysis of known mutation		One known mutation	Gene name and mutation name	Sequencing/fragment analysis/Realtime PCR
Analysis of known mutation		Mutation analysis (mutation panel)	Name for gene(s) and mutation(s)	Sequencing/fragment analysis/Realtime PCR

First version of the coding system

Published in January 2013

- A mixture of different strategies for coding of genetic analyses
 - Our suggestion
 - NPU-system
 - Everybody else's system.....

Three types of codes in NPU

- 1. Code for analysis of one gene
 - NPU19039 CFTR-gene
- 2. Code for analysis of a specific variant
 - NPU19280 CFTR-gene (5T VARIANT) DNA(spec.)—
CFTR-gen(MIM602421.0086); number (0 1 2) = ?
- 3. Gene panel
 - NPU22159 HBOC-related genes group;DNA
DNA(spec.)—Hereditary breast and ovarian cancer-
related genes; seq.var.(list)

Current project under IUPAC

- Develop NPU-codes for molecular and cytogenetic analyses
- 2014-2016
- Ulla Magdal Petersen (Copenhagen)
 - Ariadna Padro i Miquel (Barcelona)
 - Xavier Fuentes-Arderiu (Barcelona)
 - Johan den Dunnen (Leiden)
 - Graham Taylor (Melbourne)
 - Jens Michael Hertz (Odense).