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RUSSIAN FEDERATION
Lomonosov Moscow State University
Legal Faculty,
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**Forensic research
of the DNA markers
of hereditary disorders**

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SAN-ANTONIO - 2014

Human Genome Project



Impacting
many
disciplines

HUMAN GENOME
GCCAAAGTATACTA
TTTCAGCCAACATC
ATCTCCACTCTCTA
AACGAGGGAATA
ATCTGTATGTATG
AAGGGAAAAA

Global Carbon Cycles
Industrial Resources • Bioremediation
Evolutionary Biology • Biofuels • Agriculture • **Forensics**
Molecular and Nuclear Medicine • Health Risks

Objectives - to consider:

1.

- The legal aspects of the forensic study of health-related information

2.

- The anticipated value of such research for the crime investigation



The legal aspects of the forensic DNA examination of medical traits

In medical molecular genetics:



International Declaration on Human Genetic Data. 16 October 2003

Recommendation N. R (92) 3 of the Committee of Ministers to Member States on genetic testing and screening for health purposes

In forensics:



The tendency is refusal to study health-related regions and even coding DNA at all.

In a number of European national legislations were introduced limitations of the scope of forensic DNA examinations, which prevent study of health-related regions.

The Netherlands

The Amendment of 2003 to the Dutch Code of Criminal Procedure allowed to derive phenotypical information from DNA found at a crime scene and belonging to an unknown suspect within strict framework only, excluding the study of medical characteristic.



Belgium

The only purpose a forensic DNA investigation may serve is the comparison of DNA profiles in order to identify persons involved in criminal behavior.

It is a criminal offense to conduct a DNA investigation for other purposes.



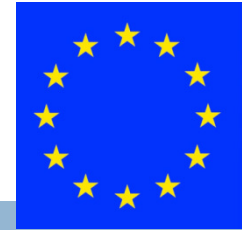
Germany

Law molecular and genetic examinations are allowed only to establish descent or to ascertain whether traces found originate from the accused or the aggrieved person.

Findings on facts other than those shall not be made; examinations designed to establish such facts shall be inadmissible.



Resolution of the Council of Europe of 30 November 2009 on the exchange of DNA analysis results (1)



When exchanging the DNA analysis results, Member States are urged to limit the DNA analysis results to chromosome zones containing **no genetic expression**, i.e. not known to provide information about specific **hereditary characteristics**.


The DNA markers of ESS are not known to contain information about specific hereditary characteristics.

Annex 1:

The European Standard Set (ESS) comprises the following DNA markers:

D3S1358
VWA
D8S1179
D21S11
D18S51
HUMTH01
FGA
D1S1656
D2S441
D10S1248
D12S391
D22S1045

Resolution of the Council of Europe of 30 November 2009 on the exchange of DNA analysis results (2)



Should science develop in such a way that it can be determined that any of the DNA markers recommended in this Resolution provide information on specific hereditary characteristics, Member States are advised to **no longer use that marker** when exchanging DNA analysis results.

Member States are also advised to be prepared **to delete any DNA analysis results**, which they may have received, if those DNA analysis results should prove to contain information on specific hereditary characteristics.

Most of countries: neither permitted nor prohibited



The legal regulation usually deals with only the traditional STR typing with no mention and thus no ban of any other forensic research.

This means that the study of the coding DNA (any region), if necessary, can be carried out within the existing legal framework.

Even if restrictions are still there, but they relate only to the activities within DNA database.

Thus, beyond this area there may be conducted also other studies.

Russia

In Federal law «On the state genome registration in Russian Federation» (2009) the genome information is specified as “personal data including the encoded information about certain fragments of deoxyribonucleic acid, not describing their physiological characteristics”.

Of the content of the law it is clear that it applies only to the activities within the framework of the DNA database.

Beyond this area there may be conducted also other studies.




The ambiguity creates legal uncertainty with respect to such a sensitive issue

Given the vulnerability of the medical traits research, even if their forensic examination is formally possible, it should be legally regulated in order to exclude any uncertainty:

- (a) if such studies are unacceptable, they should just be prohibited;
- (b) if they are acceptable, they should be allowed;
- (c) if they are acceptable with reservations, the boundaries should be defined.



- 
- Although the decision whether to use health-related DNA information in forensic research would be made on the national level, it deems vital that forensic experts and legal community should work out the common approach to this issue.



The anticipated value of the research of
medical traits for the crime
investigation

Possibilities of forensic use of health-related information



1. Revealing DNA markers of the disorders and prediction of its signs can be used while the creation for the search aims of the phenotypical portrait of the **perpetrator**.
2. In the same way the information may be used for the search of **the unknown person** whose remains were found.

Possibilities of forensic use of health-related information

3. The person of interest can be searched within specific contingents through medical records, data on uptake in certain medical institutions, etc.

In such cases it may be enough the disorder to be diagnosed by laboratory tests, even without the manifestation of the disease.



Possibilities of forensic use of health-related information

4. Beyond DNA analysis:

Signs of the hereditary diseases can be used for the search of the **perpetrator** through reports of witnesses (victims) who can denote the characteristic features of the offender.



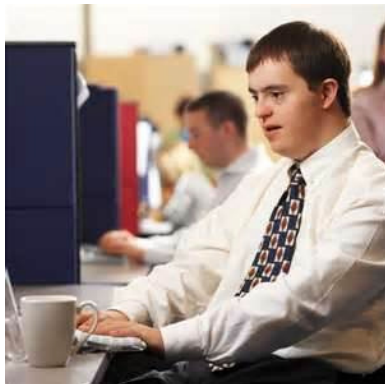
Hereditary diseases can be characterized by a specific phenotype:



- a number of external and inner defects
- expressed particular facial features,
- low or high growth (also in combination with the wrong build),
- a hoarse voice,
- hearing loss,
- mental retardation

Chromosomal disorders

Down syndrome



- Flat face
- Mongoloid eye shape
- Small ears
- Open mouth
- Mental deficiency

<http://www.yahoo.com>

Monogenic disorders



Of forensic value may be mainly DNA markers of the monogenic disorders:

- with complete or high penetrance,
- dominant
- recessive (homozygous)

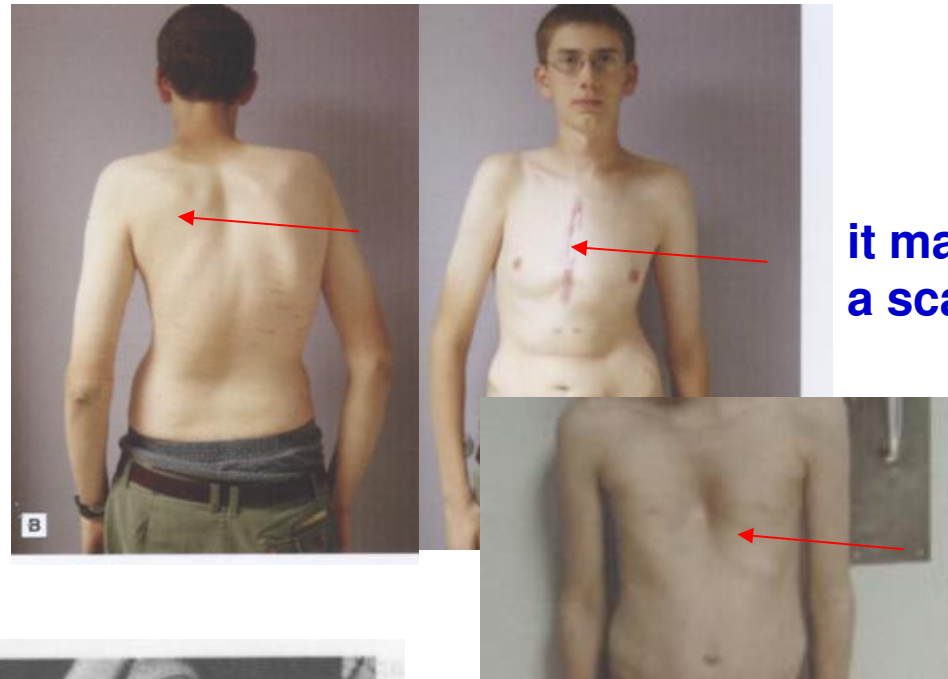
Mutation detection rate - up to 90–100% for some diseases

Can be identified by direct DNA diagnostics

Of no forensic value (at least, for the search of criminals) are severe syndromes resulting in early hard disability or early death.

Marfan syndrome

- high height
- long, slender limbs and fingers
- poorly developed subcutaneous tissue
- kyphoscoliosis
- funnel or pigeon chest deformation
- narrow, "bird" face



it may be
a scar



long thin fingers
(arachnodactyly)

K.L. Jones, 2011

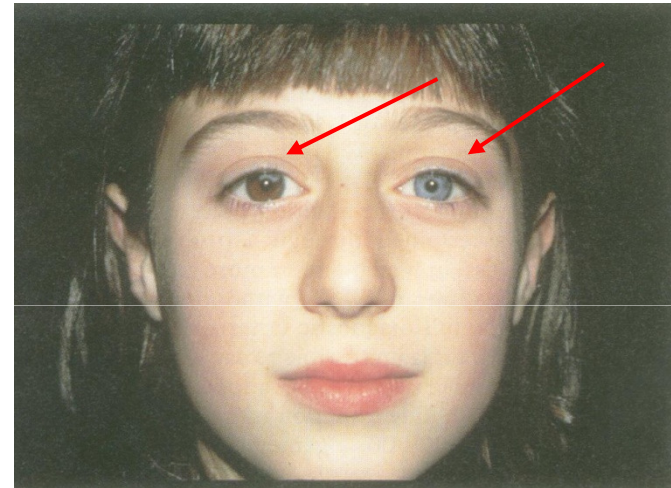
Бочков Н.П. и др., 2011

Waardenburg syndrome

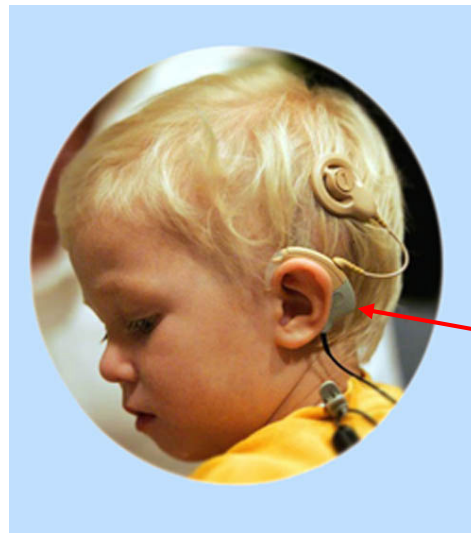


White strand

Broad bridge



**Changes in pigmentation of the iris
(heterochromia)**



Hearing deficiency

Read A.P., Newton V.E., 1997

Van der Woude syndrome

Cleft upper lip
and palate

Dimples on the
lower lip



Copyright, 2004.
Dr. Santiago Díaz Rentería

Marshall syndrome

- Wide-set eyes (hypertelorism)
- Short upturned nose
- Sunken nose
- Open mouth
- Prominent forehead
- Hearing loss



Козлова С.И., Демикова Н.С., 2007



К.Л. Jones, 2011

Williams syndrome

(“Sd of Elf face“, “Sd of unusual face”)

- epikant,
- a short nose with open nostrils forward,
- flattened midface,
- broad maxilla
- full cheeks and lips,
- a small mandible,
- open mouth,
- protruding ears,
- microcephaly (reduce the skull),
- short eye slits,
- an overbite
- a hoarse voice, caused by abnormalities or paralysis of the vocal cords.



<http://images.search.yahoo.com>

Lenz syndrome



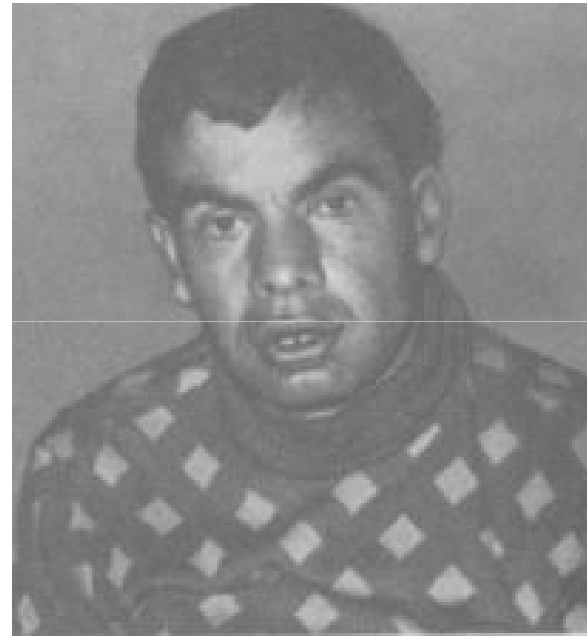
- ❑ Microphthalmia
- ❑ Protruding ears
- ❑ Mental deficiency

K.L. Jones, 2011

Mannosidose: coarse facial features

Craniofacial abnormalities:

- large skull
- prominent forehead and lower jaw
- sunken nose,
- sparse teeth,
- protruding ears



Козлова С.И., Демикова Н.С., 2007

Skin disorders: **Vitiligo**

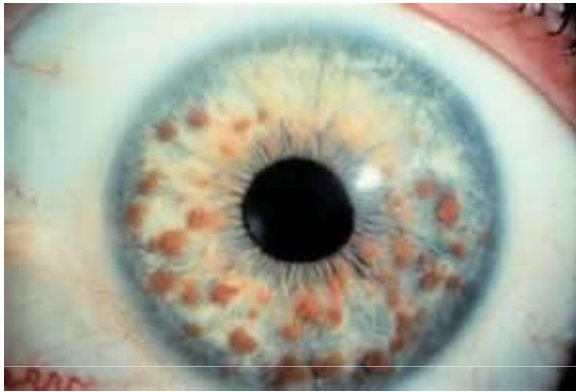


- Well-demarcated areas of depigmentation with normal skin
- 1:100

<http://www.antivitiligooil.com>

<http://www.clinicaleger.com.br>

Neurofibromatosis



Neurofibromas
(cutaneous and subcutaneous
tumors)



<http://images.search.yahoo.com>

Tuberous sclerosis



- angiofibroma cheeks as a "butterfly"
- shagreen skin thickening
- "coffee stains"



<http://images.search.yahoo.com>

Diseases that manifest at a late age

- May be relevant to the crimes committed in past when the person was clinically healthy - cold cases.
- A person might commit a crime and leave the traces in the period when the disease was not yet manifested and he was not disabled.
- E.g. **Huntington's chorea**, very heavy neurodegenerative disease, - typically characterized by the debut on the 4-6th decade of life.
- As HC has complete penetrance, its manifestation in future is actually inevitable and this can be a vector of the search in future.

Polygenic, multifactorial diseases

more common than
monogenic
disorders

rather low
mutation detection
rate

The prospects of the detection of DNA markers of the diseases

The prognostic value of the detection of DNA markers of both monogenic and multifactorial diseases will all increase:

- as more is learned about their molecular genetic nature,
- with the development of methods, especially with advent of NGS.

Reducing the price of sequencing makes the problem of sequencing solvable to practice.

The urgent task: the interpretation of the sequencing results!

Methods



- Yet NGS does not replace completely the already known methods of the DNA diagnostics.
- The choice of the technique to be used for the specific study depends on the aim of this study.
- When we have assumptions about a particular disease, then we choose the method that is most suitable and simplest for the study of the DNA marker of this disease.

Research of the health-related regions would require the development of the method



This includes:

- the choice of the disorders to be examined,
- common algorithm of examination
- particular techniques.

This suggests a big research program.

Conclusion



- In spite of a great number of problems the research of health-related DNA regions is of high forensic value and is able to add new dimensions in investigating crimes.
- The informational content of such studies will all increase with enrichment of knowledge about the molecular-genetic nature of diseases.
- The development of this area requires the formation of the legal position with regard to this kind of research.
- Legal issues should be settled in such a way as to achieve reasonable balance between interests of law enforcement and civil rights.

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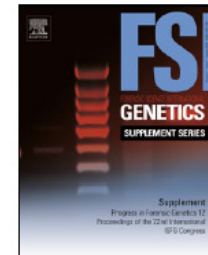


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Legislative framework and value of the forensic DNA examination of health-related information for crime investigation

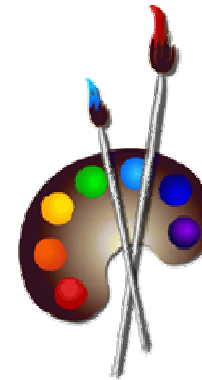


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