INTEGRATIVE APPROACH TO THALASSAEMIA IN BULGARIA

13-14 June 2009

Plovdiv, Bulgaria

THALASSAEMIAS

- Group of inherited diseases, due to a reduced or total lack of synthesis of one or more globin chains of the hemoglobin molecule
- Reduced red cells survival, hemolysis and anemia

- The most frequent monogenous diseases

Depending on the type of the affected chain:
- alpha – thalassaemic syndrome
- beta – thalassaemic syndrome
- other (rare) forms of thalassaemia

Depending on the severity:
- thalassaemia major – severe form
- thalassaemia intermedia – intermediate severity
- thalassaemia minor – mild form
- silent form – no symptoms

The most important medical and social significance is the severe form of beta thalassaemia –

Homozygous beta thalassaemia, thalassaemia major or Cooley's anaemia.

EPIDEMIOLOGY OF BETA THALASSAEMIA

- in almost all ethnic groups and geographic regions in the world
- with the highest frequency – in the countries in the Mediterranean region and around the Equator in Asia and Africa
- „thalassaemia zone" – from the Mediterranean coast through the Arab peninsula, Turkey, Iran, India and Southeastern Asia, including Thailand, Cambodia and the southern parts of China

Weatherall D, Akinyanju O, Fucharoen S, Olivieri N, and Musgrove P, "Inherited Disorders of Hemoglobin." 2006. *Disease Control Priorities in Developing Countries (2nd Edition), ed.*, 663-680. New York: Oxford University Press. DOI: 10.1596/978-0-821-36179-5/Chpt-34, permission pending

EPIDEMIOLOGY OF BETA THALASSAEMIA

CARRIER RATE OF BETA THALASSAEMIA (%)

Region	β-thal
Subsahara (Africa)	0-12
Southeastern Asia	0-11
America	0-3
Eastern Mediterranean	2-18
Cyprus	*16*
Sardinia	*20-30*
Greece	*8*
Italy	*3.7*
Macedonia	*2.9*
Turkey	*2.1*
Bulgaria	*2.4*
Spain	*0.5*
France	*0.1*

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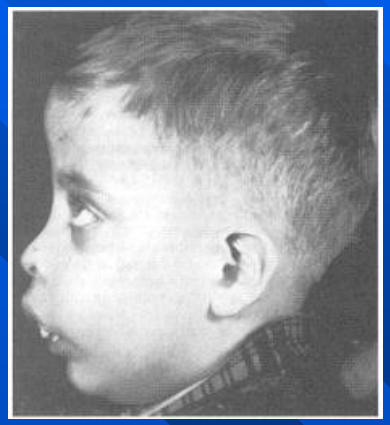
EPIDEMIOLOGY OF BETA THALASSAEMIA IN BULGARIA

- Carriers of the beta thalassaemia gene in Bulgaria – 2.4-2.5%
  (prof. Vl. Spassov, 1984 and prof. G. Petkov, 1986)
- about 170 000 Bulgarians carriers?
- There is no registry in Bulgaria!
- Total number of patients with thalassaemia major:
  242 patients for 2008 (130 children and 112 adults) *(work group on thalassaemias)*
- Newly registered patients with thalassaemia major:
  9 children for the period 2007-2008 *(work group on thalassaemias)*

CLINICAL SIGNS IN UNTREATED PATIENTS

- Severe anaemia
- Icterus
- Enlarged liver and spleen
- Skeletal deformations
- Early death – up to 4 years of age



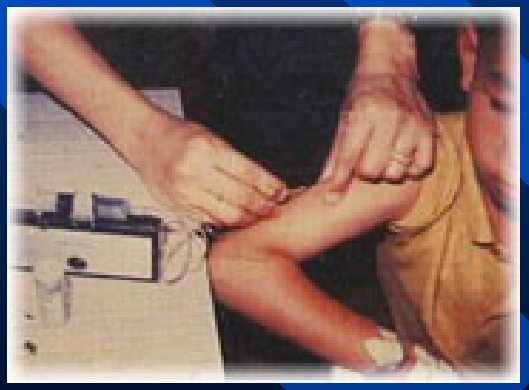
TREATMENT OF THALASSAEMIA MAJOR

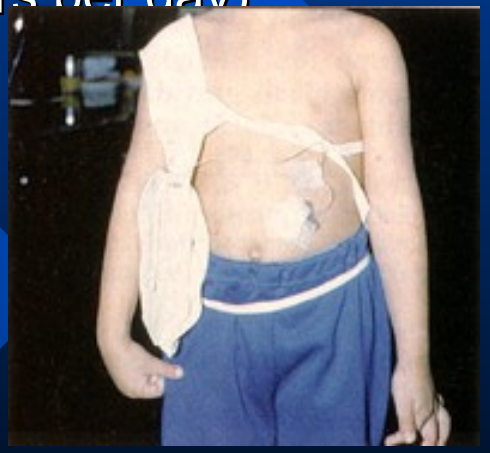
1. REGULAR TRANSFUSIONS OF RED CELLS

at intervals from 2 to 5 we

2. CONSTANT CHELATION THERAPY

1. Parenteral (*Desferal*) – prolonged subcutaneous/intravenous infusions (8-12 hours per day)





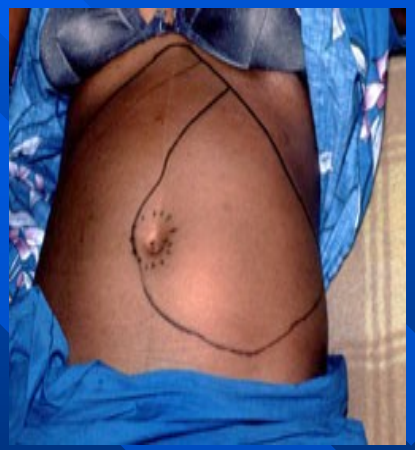
2. Oral – *Deferiprone (Ferriprox); Deferasirox (Exjade)*

3. Combined – parenteral (*Desferal*) + oral (*Ferriprox)*

COMPLICATIONS AFTER IRREGULAR TREATMENT

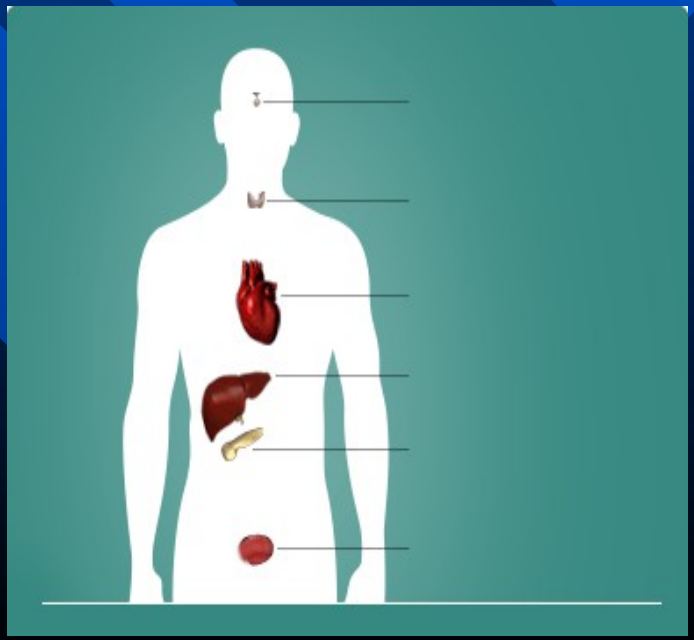
IRREGULAR TRANSFUSIONS

- delayed physical and sexual development
- skeletal deformities
- hepatosplenomegaly



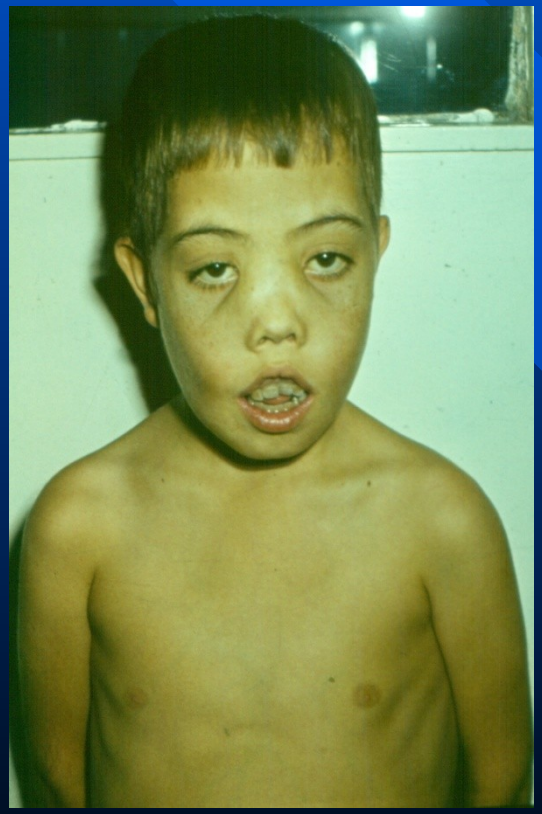
IRREGULAR CHELATION THERAPY → HAEMOSIDEROSIS

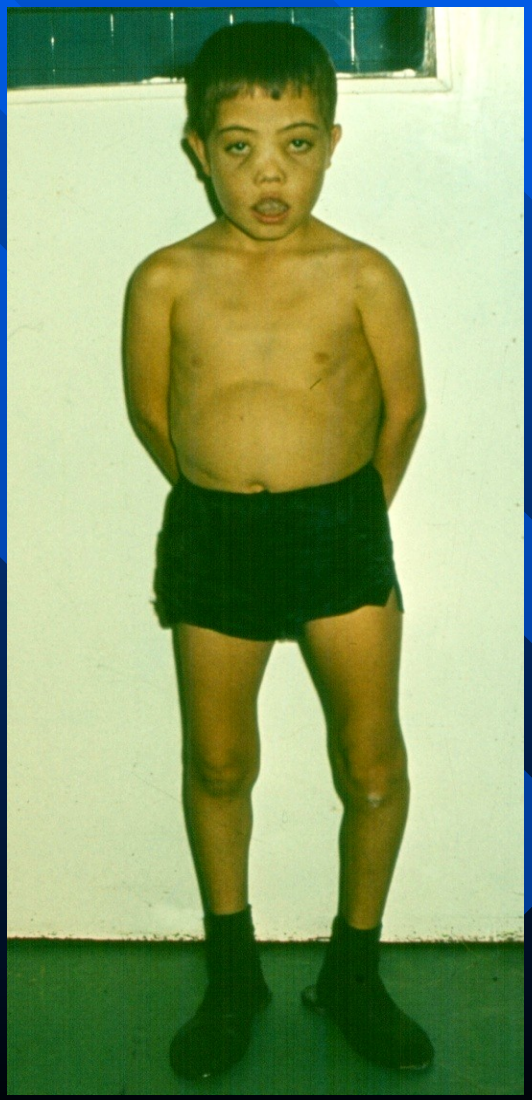
- cardiac
- endocrinology and
- liver injuries
- infectious complications
- early death – up to 30 years of age!

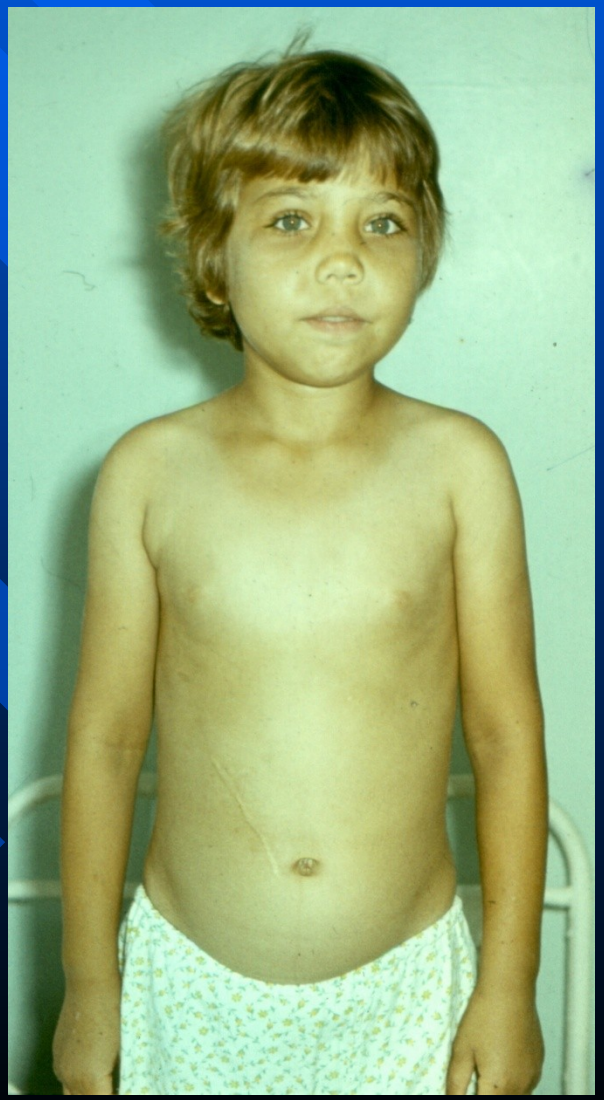


AIM OF THE CARES FOR THE PATIENTS WITH THALASSAEMIA

- warranty of adequate life
- professional realization
- creation of own families
- healthy own generation







REASONS FOR THE DEVELOPMENT OF EARLY AND SERIOUS COMPLICATIONS

COMPLEX:

- impossibility to provide for all patients adequate for their condition transfusion therapy
- lack of possibility for contemporary monitoring of the iron overload
- objective and subjective problems, connected with the carrying out of optimal iron chelating therapy

MEDICO-SOCIAL ORGANIZATION OF THE CARES FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

WORKSHOP
29.09.2007
"Sunny day", Varna

MEDICO-SOCIAL ORGANIZATION OF THE CARES FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

WORKING GROUP

FOR COORDINATION AND HELPING THE ACTIVITY, CONNECTED WITH THE MEDICO-SOCIAL ORGANIZATION OF THE CARES FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

WORKSHOP
29.09.2007
“Sunny day”, Varna

WORKING GROUP

MAIN TASKS

1. To analyze and systematize the main problems, connected with the medico-social cares for the patients with thalassaemia in Bulgaria

2. To propose to MoH a national strategy for their resolution

PROPOSALS FOR THE IMPROVEMENT OF THE QUALITY OF TREATMENT AND CARE FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

1. **Warranty of optimal transfusion treatment.**

- participation of the patients organizations in the organization of blood donation campaigns
- warranty of resources from the state/MoH for the recruitment of paid donors, particularly for patients with rare blood groups and antigen formulas
- creation of hospital blood banks in all hospitals, where transfusion therapy takes place
- the NHIC have to permit a second admission in the hospital in the frames of one month in the cases, when patients need more units of blood and more frequent transfusions

2. **Acquisition of MRI software for the investigation of heart iron overload.**

PROPOSALS FOR THE IMPROVEMENT OF THE QUALITY OF TREATMENT AND CARE FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

3. Warranty of optimal and adapted to the needs of every patient iron chelation therapy.

- change of the outdated and improper for use infusion pumps of the patients, who undergo a subcutaneous or intravenous treatment with *Desferal*
- warranty of consumables for parenteral chelation treatment (between 40 and 100 bl. per month)
- introduction in the clinical practice of the two licensed in Europe oral helators *Exjade* and *Ferriprox,* which give the possibility for choice and application of the most suitable helation program in every particular patient
- step by step increase of the quantities of the oral helators *Exjade* and *Ferriprox* with the aim most (or all) patients, suitable for only oral or combination treatment, to be included in the programs

PROPOSALS FOR THE IMPROVEMENT OF THE QUALITY OF TREATMENT AND CARE FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

4. **Creation of thalassaemia centers and interdisciplinary medical teams.**

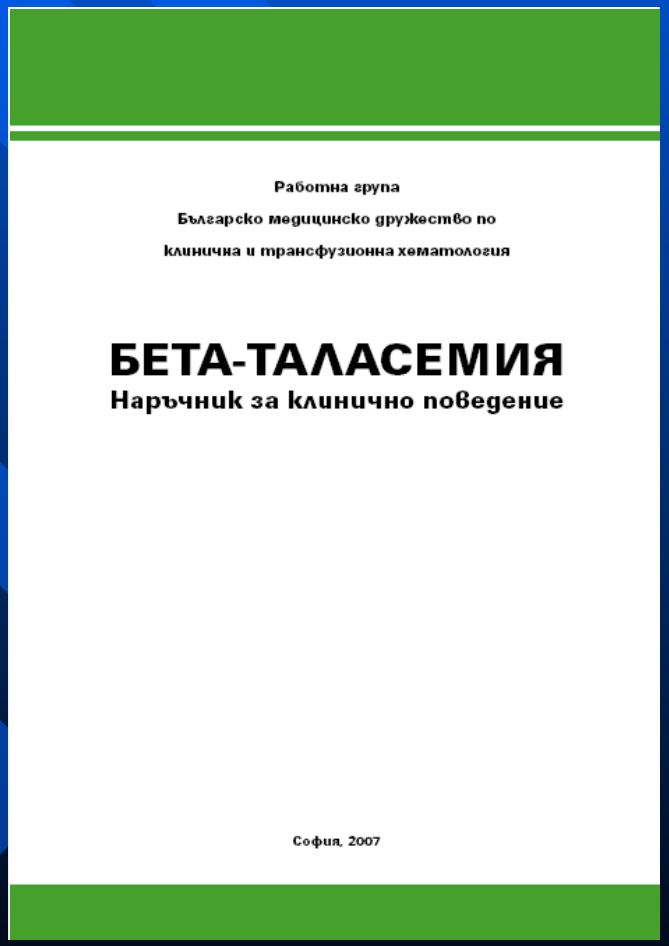
- creation of a unique (for children and adults) thalassaemia center in Sofia with a national statute
- creation of unified thalassaemia centers (on functional principle) in the university hospitals in Plovdiv, Stara Zagora, Varna and Pleven
- creation of medical teams for interdisciplinary follow up and treatment of children and adults in the thalassaemia centers, including specialists in hematology, cardiology, endocrinology, liver diseases, a psycologist and a social worker.

PROPOSALS FOR THE IMPROVEMENT OF THE QUALITY OF TREATMENT AND CARE FOR THE PATIENTS WITH THALASSAEMIA IN BULGARIA

5. Creation of standards for clinical behavior for patients with thalassaemia.
6. Creation of a national prophylaxis program.
7. Cooperation with the patients organizations and assistance of the patients for their social adaptation and realization.
8. Approval of an expert (work) group for coordination and assistance of the activity, connected with the medicosocial organization of the cares for the patients with thalassaemia in Bulgaria

WORK GROUP

- Edition of a *Handbook* for clinical behavior in *beta thalassaemia*.
- Creation of a preliminary register and initial data base for the patients with thalassaemia in Bulgaria.
- Participation in a National meeting in MoH on the problems of the treatment of the patients with thalassaemia in Bulgaria.
- Development of criteria for the treatment with the oral helator *Exjade*.
- Development of a protocal for the treatment with *Exjade.*
- Participation in the project частие в проекта “National register of the patients with thalassaemia in Bulgaria”.



WORK GROUP

- Development of a project for the creation of interdisciplinary teams for the treatment and follow up of children and adults with thalassaemia in the university hospital structures.
- Development of a project: *Recommendations for good practice for children and adults with thalassaemia in Bulgaria.*
- Development of a project for the creation of a hospital blood bank in MHAT "St. Marina" Varna.
- Development of a project for the investigation of heart iron overload by T2*MRI in Sofia and Varna.
- Participation in national and international scientific forums and activities of the patients organizations.