AZIENDA OSPEDALIERA UNIVERSITARIA INTEGRATA di VERONA
(D.Lgs. n. 517/1999 - Art. 3 L.R.Veneto n. 18/2009)
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INFORMED CONSENT for NEONATAL SCREENING DISCLOSURE for the PROCESSING of GENETIC DATA

Dear Parents,

the Veneto Region has progressively introduced some neonatal screenings aimed at early identification of infants with congenital hypothyroidism, phenylketonuria, cystic fibrosis, galactosemia, biotinidase deficit, glucose-6-phosphate dehydrogenase deficiency and congenital adrenal hyperplasia. It is a non-prescription charge performance and therefore free, for which an information brochure is distributed at the Maternity Ward. In particular, screenings for congenital hypothyroidism, phenylketonuria and cystic fibrosis, established by national legislation, are mandatory[1] and do not require obtaining informed consent from parents.

In addition to long-running active programs, from 1st January 2014, the Veneto Region has also introduced expanded metabolic screening.[2] This new service, also totally at the Region's expense, will identify in time for a truly effective therapy the infants with the following diseases: Maple syrup urine disease or branched-chain ketoaciduria (MSUD), Tyrosinemia type I and II (TYR-TYR-I and II), glutaric acidemia type I (GA-I), isovaleric acidemia (IVA), propionic acidemia (PA), methylmalonic acidemia (MMA), cobalamin deficiency (type A, B, C and D), 3-hydroxy-3-methylglutaryl-CoA lyase (HMG) deficiency, 3-metilcrotonil-CoA carboxylase (3MCC) deficiency, Carnitine Carrier urinary deficiency (CUD), carnitine palmitoyltransferase I deficiency (CPT-I), carnitine palmitoyltransferase II deficiency (CPT-II), Carnitine-acylcarnitine Translocase deficiency (CACT), medium chain acyl-CoA dehydrogenase deficiency (MCAD) very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD), multiple acyl-CoA dehydrogenase deficiency (GA-II).

Both in the case of neonatal screening required by law and for the additional ones, these diseases are present from birth and if not treated early can have damaging effects on a neurological, cardiac, muscle, lung level, and to other organs. In severe cases these can lead to premature death. Early identification of these infants and prompt therapy implementation can help prevent much of the symptomatology, ensuring the child has a good quality of life, or, in more severe forms, an important slowing down of the disease progression.

We ask your consent to carry out additional neo-natal screening by filling out the attached form.

With your consent, the analysis will be carried out by utilising the same drops of blood collected in order to perform the screening required by law, without any need for further collections.

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1 Law 5th February 1992 n. 104, Law 23rd December 1993 n. 548.
2 Regional Law 1308/13.

Should the screening show the possibility of one of the listed diseases, within twenty days you will be contacted by a doctor who will schedule a check for further diagnostic tests, the outcome of which will clarify whether the disease is actually present. If the suspicion is confirmed, appropriate treatment will be started immediately, allowing better control of the disease.

Within the context of some new-born screenings, both mandatory and additional, a genetic analysis on the same drop of blood taken at birth may need to be carried out. In the unlikely event that this analysis should be necessary, **we ask now for your consent to the genetic analysis**. Should the genetic analysis prove to be positive and **you declared you wish to know the results**, you will be offered genetic consultation.

Should a genetic analysis be performed, **your consent is also required for the treatment and storage of personal and sensitive data**. In the absence of such consent, it will not be possible to perform the genetic analysis should this be necessary. Only some personal and family information is requested, strictly necessary for the tests.[3] You can in any case and at any time, find out all the data concerning your child, know how this was acquired, check whether it is accurate, complete and up-to-date, as well as enforce your rights in this regard[4,5]

After running all the analysis, the remaining sample will be stored for three years in anonymous form.[6] **You are being asked to consent to its preservation for a further 20 years. During these years**, the sample could be used, in anonymous form,[7] to usefully investigate the diagnosis or therapy of the disease or, in the case of a healthy new-born, for research for other diseases (e.g., it could be part of a "control" group, that is a set of healthy people, with which to compare individuals with a particular disease).

The data resulting from the screening test, both electronic and paper form, will be used exclusively for the screening and stored at the University Hospital of Verona so as to minimise, by adopting suitable and preventive safety measures, the risk of destruction and accidental loss, unauthorised access, treatment which is not allowed or not consistent with the purpose of the collection, in compliance with professional secrecy. The test result will be communicated to the staff of the maternity ward where your child was born (generally consisting of medical personnel) or to you (or to your delegate with a written proxy, to whom it will be delivered in a closed envelope), and, without any reference for identifying the individual child, to the regional bodies responsible for the matter (Rare Disease registry, Department of Health). Other health care facilities that perform, on behalf of AOUI Verona, contextual biochemical investigations to the screening procedures may become informed of this data in their capacity as data controllers.

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[3] In accordance with Authorisation n. 8/2012 - General authorisation for the processing of genetic data - December 13th, 2012, by the Authority for the Protection of Personal Data, par. 4.3.

[4] Art. 7 of the "Privacy Code".

[5] In particular, you have the right to oppose the processing of genetic data for legitimate reasons by simply addressing yourself in writing to the Director of the Neonatal Metabolic Disease Centre of the University Hospital of Verona, Piazzale P.le L.A. Scuro, 10, Verona, phone 045.8126677, e-mail attilio.boner@ospedaleuniverona.it, who is responsible for the processing of genetic data, appointed by the Hospital University of Verona as person in charge.

[6] A numeric code is associated to the blood adsorbed on the card that, if necessary, allows the medical staff of the Neonatal Metabolic Diseases Centre of the Hospital University of Verona, to go back to the name of the new-born from whom the blood was collected.

[7] No information which makes it possible to trace the name of the new-born from whom the blood was collected is associated to the blood adsorbed on the card.

The same data will be stored at the Neonatal Metabolic Disease Centre of the University Hospital of Verona in accordance with existing regulatory obligations concerning the conservation of diagnostic documents.[8] The same results will not be disclosed to other parties, unless they are essential for the health protection of / the new-born or of a family member, on the basis of a specific request substantiated by appropriate health records,

I hope that this initiative will find your consent and I would like to thank you for your collaboration

Prof. Attilio Boner

[8] The Directive of the Ministry of Health no. 61 of 19 December 1986 which provides for the retention of diagnostic laboratory records for 20 years is currently in force.

The undersigned _____

(full name of a parent or other person named below)

Born in _____on ___/___/____

resident in _____ Post Code _____ Address _____

phone _____ mobile _____

as _____

(parent, or family member, or person in charge of the maternity ward, or other person indicated by art. 82, paragraph 2, letter. a) of the Privacy Code)

of

_____

(name, surname of the new-born)

born in_____on ___/___/_____

DECLARES

that she/he was correctly informed in relation to the neonatal screening programs and to have read the information concerning the processing of personal data for the purposes connected with said screenings (see document "INFORMED CONSENT for NEONATAL SCREENING - DISCLOSURE for the PROCESSING OF SENSITIVE GENETIC DATA").

AND AGREES CONCERNING THE CHILD

□Yes□No **to the performance of the additional new-born screening;**

□Yes□No **to the eventual execution of the genetic analysis if needed in the context of both mandatory and additional screening;**

□Yes□No **to the processing of sensitive personal data according to current legislation should the genetic analysis be performed, in accordance with the information given (Legislative Decree. N. 196/2003);**

□Yes□No **to the conservation for further 20 years in anonymous form of the residue biological material for any investigations that in the future could be indicated for the diagnosis / therapy of the disease and / or for study or research, also relating to diseases other than those for which consent to the data processing was sought.**

DECLARES

□Yes□No **that he/she wishes to know the results of the genetic tests carried out in case of need should the new-born's screening be doubtful. You will have the right to find out the result of this analysis even if you had stated you did not wish to know, should you change your mind in the meantime.**

Signature *(parent / other person indicated above)* _____

The doctor who collected the consent:

Dr._____ Signature (Doctor) _____

Place and date: _____