APPENDIX

Appendix 1

KOMISI ETIK PENELITIAN KESEHATAN (KEPK)
FAKULTAS KEDOKTERAN UNIVERSITAS DIPONEGORO
DAN RSUP dr KARIADI SEMARANG
Sekretariat : Kantor Dekanat Lt.3 FK Undip
Jl. Dr. Sutomo 18. Semarang
Telp.024-8311523/Fax. 024-8446905

RSUP Dr. KARIADI

ETHICAL CLEARANCE

No 17 /EC/FK/RSDK/2010

Komisi Etik Penelitian Kesehatan Fakultas Kedokteran Universitas Diponegoro/ RSUP. dr. Kariadi Semarang, setelah membaca dan menelaah USULAN Penelitian dengan judul :

IDENTIFICATION OF GENETIC CAUSES OF RETINITIS PIGMENTOSA IN THE INDONESIAN POPULATION USING HIGH RESOLUTION HOMOZYGOSITY MAPPING

Peneliti Utama : dr. A. Kentar Arimadyo Sulakso, Sp.M

Penelitian : Dilaksanakan di St.Radboud University di Nijmegen Netherlands dan Laboratorium CEBIOR FK UNDIP Semarang

Setuju untuk dilaksanakan, dengan memperhatikan prinsip-prinsip yang dinyatakan dalam Deklarasi Helsinki 1975, dan Pedoman Nasional Etik Penelitian Kesehatan (PNEPK) Departemen Kesehatan RI 2004

Peneliti harus melampirkan 2 kopi lembar Informed consent yang telah disetujui dan ditandatangani oleh peserta penelitian pada laporan penelitian.

Semarang, 17 Februari 2010

Fakultas Kedokteran Undip
Dekan

dr. Soejoto, PAK, Sp KK(K)
NIP. 130 368 078

Komisi Etik Penelitian Kesehatan
Fakultas Kedokteran Undip/RSUP. Dr. Kariadi
Ketua

Prof. Dr.dr. Tjahjono, Sp PA(K)FIAC
NIP. 130 368 076

Appendix 2

CENTER FOR BIOMEDICAL RESEARCH
FACULTY OF MEDICINE DIPONEGORO UNIVERSITY

MATERIALS TRANSFER AGREEMENT

This Materials Transfer Agreement is made on this day, of 30 October 2009 by and between:

The Faculty of Medicine, Diponegoro University, an Indonesian Research institution existing under the laws of the Republic of Indonesia, having its registered office at Jl. Dr. Sutomo No. 18, Semarang, Central Java Province, Indonesia (hereinafter referred to as "**First Party**"), represented by Prof. Sultana M. H. Faradz, MD, PhD, in this matter acting in her capacity as the Director of The Center For Biomedical Research, Faculty of Medicine, Diponegoro University.

Radboud University Nijmegen Medical Centre, a research institution existing under the laws of Dutch Government having its registered office at Nijmegen, The Netherlands, [Tel] +31.243614017, [Fax] +31.243668752, represented by Prof. Frans P.M. Cremers, head Division of Molecular Genetics of the Department of Human Genetics, in this matter acting in his capacity as Recipient and Scientist (hereinafter referred to as "**Recipient**");

And

dr. Kentar Arimadyo Sulakso SpM, domiciled at Jl Bukit Barisan C1 no 9, Perum Bukit Permata Puri, Ngaliyan, Semarang, Central Java Province, Indonesia, Tel: +62-24-7629003 and Dr. Rob W.J. Collin (hereinafter referred to as "**Scientists**").

(The Recipient and Scientists shall collectively hereinafter referred to as "**Second Party**")

In consideration of the Recipient's and the Scientist's covenant and premises contained herein, the First Party agrees to provide the Materials to the Second Party for the sole purpose of the study and for specific assays (Research Plan/Protocol) described in Appendix B, which shall be an integral part of this Agreement, upon the terms and conditions hereinafter appearing

1. DEFINITIONS

In this Agreement, definitions that are used are as the following meaning:

Materials : means Original Materials, Progeny, and Unmodified Derivatives of the biological specimens and or data described in Appendix A.

Original Materials : means substances as described in the Appendix A.

Progeny : unmodified genetic descendant from the Materials.

Unmodified Derivatives : substances created by the Recipient which constitute an unmodified functional subunit or product expressed by the Original Materials.

Modifications : substances created by the Recipient which contain and/or incorporate the Materials.

Research Plan/Protocol : study and for specific assays and research to be under taken as described in the Appendix B.

2. OWNERSHIP OF MATERIALS

The Second Party acknowledges that rights, title and interest of the Original Materials are the property of the First Party and the First Party shall retain ownership and the Modifications.

3. USE OF MATERIALS

The Second Party undertakes to use the Materials and Modifications, solely for the purpose of Research Plan/Protocol as further described in the Appendix B (The research Proposal); and in accordance with the terms of this Agreement and laws, and regulation. The Second Party will not undertake to transfer, distribute, release, or disclose by any means, either intentional or accidental, the Materials or Modifications except as expressly stated in Appendix B for the sole purpose of the Research Plan/Protocol under the supervision of the Scientist; and also not to use the Materials or Modifications for any purpose other than as expressly stated in Appendix B/non-commercial research.

4. THE RESEARCH PLAN/ PROTOCOL

The Research Plan/Protocol shall be developed together by the Parties in providing best effort to conduct the research, the tests, and the experiments related to the Research Plan/Protocol within the jurisdiction of the Republic of Indonesia The Second party shall send in confidence, to the First Party any and all data, records, and results derived from the Materials and Research Plan, including detailed records of direct use of the Materials.

5. INTELLECTUAL PROPERTY RIGHTS

The Second Party acknowledges that the Materials or Modifications are or maybe the subject of patent application. Nothing in this Agreement grants any implied or express license or right under any patents or in any know-how or trade secrets other proprietary rights to use the Materials or Modifications or any product or process related thereto for profit-making or commercial purposes, including but not limited to, production, sale, screening or drug design. The Recipient agrees to negotiate in good faith a license with the First Party prior to making any such profit-making or commercial use. The First Party shall have no obligation to grant such license to the Recipient, and may grant exclusive or non-exclusive licenses to others who may be investigating uses of the Materials or Modifications.

6. RETURN OF MATERIALS AND MODIFICATIONS

The First Party may request to the Second Party to return any and all unused Materials, Modifications and all of the data, records, and results derived from the Materials and Research Plan/Protocol.

7. PUBLICATION

The use of any data, results, or concepts (hereinafter referred to as "**Outputs**"), derived from use of the Materials in presentations, abstracts, publications (both peer-reviewed and not peer-reviewed), grants, or other means of disseminations by the Recipient and/or Scientist shall require written consent from the First Party. In the event that the Second Party wishes to use Outputs for dissemination of any kind as described above, the Second Party shall provide a written request along with a copy of the presentation, abstract, manuscript, grant or other medium to the First party prior to any requested date of dissemination. The inclusion of the First Party in the Outputs will be as author or co-author, shall be described in details in the Research Plan/Protocol. The First Party agrees that it will acknowledge the Second Party's publications, as academically and scientifically appropriate

8. CONFIDENTIALITY

The Second Party shall treat in confidence any information relating to the Materials and/or Modifications saved.

9. DISCLAIMER OF WARRANTY

The First Party makes no representations, conditions or warranties either express or implied with respect to any of the Materials or Modifications and disclaims any implied warranty, condition or representation that the Materials or Modifications. The First party shall not be liable for loss whether direct, consequential, incidental or special (and whether arising out of contract or tort) which the Recipient or the Scientist may suffer arising from the use, handling, storage, defect, error, fault or failure to perform with respect to the Materials or Modifications.

10. INDEMNITY

The Second Party hereby jointly and severally agree and undertake to indemnify, hold harmless and defend the First Party against any and all claims, actions, damages, liabilities, loss whatsoever (including all legal costs and expenses on a full indemnity basis) arising out of or resulting from directly, the possession, use and/or storage of any of the Materials and Modifications or by reason of any breach of the terms herein by the Recipient and/or the Scientist. The First Party should be liable for consequential or incidental damages arising from breach or breaches of this Agreement. No action, whether in contract or tort (including negligence) or otherwise arising out of or in connection with this Agreement may be brought by the Recipient or the Scientist more than 12 months after the cause of action has occurred.

11. TERMINATION

This Agreement will terminate on the earliest of the following date: (a) on the completion of the implementation activities set forth in the Research Plan/Protocol as described in the Appendix B, or (b) on 30 (thirty) days' written notice by either party to another. The First Party may terminate this agreement if it is of the view that the Recipient and/or the Scientist

are in breach of any of the terms here of and such breach, if capable of being remedied, is not remedied by the Recipient or Scientist after 30 (thirty) day's Notice by the First Party.

12. ARBITRATION

12.1 Failing such an amicable settlement, any and all disputes, controversies, and conflicts arising out of, or in connection with this Agreement, or its performance, shall be finally settled by arbitration in accordance with the Arbitration Rule of the International Chamber of Commerce ("**ICC**"), which rules are deemed to be incorporated by reference into this clause. The Arbitration proceedings shall take place in Jakarta and shall be conducted in English.

12.2 The Parties agree that the Panel of Arbitrators shall consist of 3 (three) arbitrators. The First Party and the Second Party shall respectively have the right to appoint 1 (one) arbitrator and should one party fail to appoint its arbitrator in 14 (fourteen) days from the appointment of the first arbitrator, then such arbitrator shall be appointed by the ICC. The 2 (two) arbitrators so appointed shall jointly appoint the third arbitrator who will act as the Chairman of the Panel of Arbitrators. Should the 2 (two) arbitrators fail to appoint the third arbitrator in 14 (fourteen) days from the appointment of the second arbitrator, then such third arbitrator shall be appointed by the ICC.

13. NOTICE

First Party: Prof. Dr. Sultana M.H. Faradz, MD, PhD, Center for Biomedical Research, Faculty of Medicine, Diponegoro University
[Address]: Jl. Dr. Sutomo, No 14 Semarang, Central Java Province, Indonesia
[Fax:] +62-24-8454714

Second Party:

The Recipient and Scientist:
Prof. Dr. Frans P.M. Cremers, Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands,
[Address]: Geert Grooteplein Zuid 8, 6525 GA Nijmegen
[Tel] + 31.243614017
[Fax] +31.243668752

The Scientist: Dr. Kentar Arimadyo Sulakso,SpM
[address] : Jl. Dr. Sutomo, No 14 Semarang, Central Java Province, Indonesia
[Fax:] : +62-24-8442216

13. GOVERNING LAWS

This Agreement shall be governed, interpreted and applied in accordance with the laws of the Republic of Indonesia.

First Party,

Institutional Representative

Prof. Dr. Sultana M.H. Faradz, PhD
Director of Center For Biomedical Research, Faculty of Medicine, Diponegoro University, Indonesia

Date: 3/11/09

Second Party,

Recipient Institutional Representative:

Prof. Dr. Frans P.M. Cremers
Department of Human Genetics
Radboud University Nijmegen Medical Centre, Geert Grooteplein Zuid 8 , 6525 GA Nijmegen, The Netherlands

Date: 30 October 2009

Scientist: dr. Kentar Arimadyo, SpM

Date: 30 October 2009

Approved by,

A/Prof. Soejoto, MD, DV
Dean of Faculty of Medicine, Diponegoro University

Date: 4 November 2009

Appendix 3

Concentration of DNA control panel

No	Ind No	SEX	No UMCN	Conc	No	Ind No	SEX	No UMCN	Conc
1	01/DS	M	054060	351.5	104	01/Pras/05	V	054327	470
2	02/HR	M	054061	146.9	105	02/Pras/05	V	054328	1006
3	03/D	M	054062	266.2	106	03/Pras/05	V	054329	305
4	04/AW	V	054063	178.5	107	04/Pras/05	V	054330	153
5	05/TJ	V	054064	739.3	108	05/Pras/05	V	054331	274
6	06/INF	V	054065	199.7	109	06/Pras/05	V	054332	466
7	07/M	M	054066	393.6	110	07/Pras/05	V	054333	186
8	08/SP	M	054067	667.8	111	08/Pras/05	V	054334	653
9	09/SG	M	054068	509.7	112	09/Pras/05	V	054335	101
10	10/ST	M	054069	670.6	113	10/Pras/05	V	054336	80.6
11	11/AR	M	054070	357.2	114	42/Stock/05	V	054337	519
12	12/MJ	M	054071	643	115	43/Stock/05	V	054338	518

13	13/JN	M	054072	478	116	45/Stock/05	V	054339	620
14	14/FY	M	054073	132.6	117	14/Stock/05	M	054340	238
15	15/NN	V	054074	88.15	118	47/Stock/05	V	054341	354
16	16/ARF	M	054075	408.5	119	48/Stock/05	M	054342	789
17	17/SW	M	054076	316.1	120	49/Stock/05	M	054343	604
18	18/NG	V	054077	353	121	50/Stock/05	M	054344	487
19	19/YK	M	054078	519.4	122	53/Stock/05	M	054345	726
20	20/WN	M	054079	807.5	123	54/Stock/05	V	054346	687
21	21/F	V	054080	129.9	124	55/Stock/05	V	054347	367
22	22/LW	M	054081	360.4	125	56/Stock/05	V	054348	264
23	23/B	M	054082	467	126	57/Stock/05	V	054349	406
24	24/ABD	M	054083	369.1	127	58/Stock/05	V	054350	580
25	25/AM	M	054084	366.3	128	59/Stock/05	V	054351	178
26	26/SB	M	054085	301.2	129	61Stock/05	V	054352	281
27	27/SK	V	054086	242.2	130	64/Stock/05	V	054353	186
28	28/WR	V	054087	342.3	131	65/Stock/05	V	054354	362
29	29/TH	M	054088	136.2	132	67/Stock/05	V	054355	266
30	30/HL	V	054089	603.2	133	68/Stock/05	V	054356	212
31	31/BH	M	054090	278.9	134	69/Stock/05	V	054357	369
32	32/NT	M	054091	26.02	135	70/Stock/05	M	054358	169
33	33/HM	M	054092	217.3	136	71/Stock/05	M	054359	365
34	34/KS	M	054093	449	137	72/Stock/05	M	054360	254
35	35/RB	V	054094	325.1	138	73/Stock/05	M	054361	782
36	36/MR	V	054095	434.3	139	74/Stock/05	M	054362	140
37	37/SR	V	054096	45.3	140	75/Stock/05	M	054363	532
38	38/HK	V	054097	265	141	01/Stock/05	M	054364	1251
39	39/SJ	V	054098	93.67	142	02/Stock/05	V	054365	334
40	40/SBD	M	054099	324.9	143	03/Stock/05	V	054366	435
41	41/SKR	V	054100	130	144	04/Stock/05	V	054367	568
42	42/RT	V	054101	389.2	145	05/Stock/05	M	054368	216
43	43/DM	V	054102	448.3	146	17/Stock/05	M	054369	674
44	44/PW	M	054103	267.2	147	21/Stock/05	M	054370	246
45	45/HS	M	054104	400.3	148	26/Stock/05	M	054371	697
46	46/ST	V	054105	450.5	149	15/Stock/05	M	054372	367
47	47/BW	M	054106	483	150	13/Stock/05	M	054373	464
48	48/AK	M	054107	229.6	151	77/Stock/05	M	054374	334
49	49/MU	V	054108	199.9	152	79/Stock/05	V	054375	483
50	50/SH	M	054109	272.6	153	81/Stock/05	V	054376	603
51	51/BP	V	054110	225.2	154	82/Stock/05	V	054377	273.8
52	52/EP	V	054111	79.22	155	84/Stock/05	V	054378	165
53	53/PSS	V	054112	130.1	156	85/Stock/05	M	054379	455
54	54/TA	V	054113	114.5	157	01/CLH/08	M	054380	656
55	55/SL	V	054114	178.9	158	02/CLH/08	M	054381	427
56	56/SI	V	054115	186	159	03/CLH/08	M	054382	705
57	57/PJ	V	054116	274.2	160	04/CLH/08	M	054383	465
58	58/SNT	V	054117	159.8	161	05/CLH/08	M	054384	416
59	59/SRJ	V	054118	193.2	162	06/CLH/08	M	054385	443
60	60/MYT	V	054119	40.97	163	07/CLH/08	M	054386	277
61	61/AYN	V	054120	195	164	08/CLH/08	M	054387	835
62	62/T	V	054285	22.3	165	09/CLH/08	M	054388	426
63	63/DU	V	054286	15.05	166	10/CLH/08	M	054389	363
64	64/SS	V	054287	344.4	167	11/CLH/08	M	054390	132
65	65/STR	V	054288	201.7	168	12/CLH/08	V	054391	247
66	66/PH	M	054289	275.6	169	13/CLH/08	M	054392	165
67	67/SNW	M	054290	383.1	170	14/CLH/08	M	054393	355
68	68/Pi	V	054291	148.8	171	15/CLH/08	M	054394	184
69	69/BSK	M	054292	289.6	172	16/CLH/08	V	054395	398
70	70/DD	M	054293	232.4	173	17/CLH/08	V	054396	135
71	71/HTT	V	054294	182.2	174	18/CLH/08	V	054397	397
72	72/ANT	V	054295	355	175	19/CLH/08	V	054398	365
73	73/PWT	M	054296	369.2	176	01/CK/09	V	054399	544
74	74/TTW	V	054297	339.4	177	02/CK/09	V	054400	550
75	75/SGT	M	054298	199.4	178	03/CK/09	V	054401	263
76	76/IDH	V	054299	193.7	179	04/CK/09	V	054402	654
77	77/ABW	M	054300	193.3	180	05/CK/09	V	054403	188
78	78/BHS	M	054301	93.81	181	06/CK/09	V	054404	338

79	79/TJO	M	054302	108.6	182	07/CK/09	V	054405	272
80	80/JRK	M	054303	147.1	183	08/CK/09	V	054406	813
81	81/SDY	M	054304	99.1	184	09/CK/09	V	054407	212
82	82/KNT	M	054305	247	185	10/CK/09	V	054408	571
83	83/THY	V	054306	236.1	186	11/CK/09	V	054409	341
84	84/HSO	M	054307	327.2	187	12/CK/09	V	054410	361
85	85/RAM	V	054308	181.3	188	13/CK/09	M	054411	624
86	86/STR	M	054309	172.6	189	14/CK/09	M	054412	527
87	87/RKD	M	054310	115.2	190	15/CK/09	M	054413	476
88	88/DSW	V	054311	76.82	191	16/CK/09	M	054414	254
89	89/UCY	V	054312	121	192	17/CK/09	M	054415	353
90	90/HBW	M	054313	100	193	18/CK/09	M	054416	318
91	91/SWN	V	054314	218.2	194	19/CK/09	M	054417	431
92	92/HDY	M	054315	262	195	20/CK/09	M	054418	279
93	93/SGT	M	054316	219.8	196	21/CK/09	V	054419	450
94	94/MKR	M	054317	232.5	197	22/CK/09	M	054420	240
95	95/FSW	V	054318	135.8	198	23/CK/09	M	054421	512
96	96/TSR	V	054319	135.9	199	24/CK/09	M	054422	281
97	97/AWN	V	054320	147.8	200	25/CK/09	V	054423	223
98	98/JRT	V	054321	347.5	201	04/KTR/08	V	054424	954
99	99/KSD	M	054322	110.5	202	05/KTR/08	M	054425	867
100	100/EDH	V	054323	77.28	203	06/KTR/09	V	054426	774
101	101/DWS	V	054324	51.57	204	07/KTR/09	V	054427	862
102	102/WWK	V	054325	47.92	205	08/KTR/10	M	054428	394
103	103/PWS	V	054326	128.6	206	09/KTR/10	M	054429	512
					207	10/KTR/11	M	054430	115

Appendix 4

JUDUL PENELITIAN **:** IDENTIFICATION OF GENETIC CAUSES OF RETINITIS PIGMENTOSA IN THE INDONESIAN POPULATION USING HIGH RESOLUTION HOMOZYGOSITY MAPPING

INSTANSI PELAKSANA **:** BAGIAN ILMU KESEHATAN MATA FK UNDIP/ RS Dr KARIADI SEMARANG DAN CEBIOR FK UNDIP

PERSETUJUAN SETELAH PENJELASAN
(INFORMED CONSENT)

Tujuan Penelitian:

Bapak/Ibu akan kami ajak untuk berpartisipasi dalam penelitian ini.

Bapak/ Ibu menderita gangguan penglihatan akibat adanya kelainan pada lapisan sel foto

reseptor retina yang berfungsi untuk penglihatan. Penyakit ini dapat mengakibatkan kebutaan total pada stadium yang lanjut. Gejala awal penderita berupa rabun senja disertai adanya penyempitan lapang pandang penderita secara perlahan. Penyempitan lapang pandang ini bermula dari daerah perifer dan pada akhirnya juga mengenai daerah sentral yang berakhir pada kebutaan total. Penyakit ini diturunkan secara genetik melalui orang tua penderita. Adapun jenis penurunan sifatnya berupa automal dominan dimana terjadinya penyakit ini diperoleh dengan cukup adanya satu copy gen autosomal yang diperoleh dari salah satu orang tuanya, autosomal resesif dimana penyakit ini diturunkan apabila didapat 2 copy gen autosomal yang termutasi dan diperoleh dari kedua orangtuanya dan X-linked apabila penyakit ini diperoleh dari mutasi gen yang diturunkan melalui kromosom sex X. Untuk kasus autosomal resesif kedua orang tua penderita mempunyai andil dalam penurunan sifat penyakit ini.

Dengan diketahuinya sifat penurunan dan mutasi gen penyebab Retinitis Pigmentosa ini penderita mampu merencanakan masa depannya secara lebih bijaksana dan mantap. Misalnya bila ingin mencari pasangan ataupun ingin memiliki anak, dimana selama ini masih ada kekhawatiran tentang kemungkinan anaknya juga ikut terkena penyakit ini. Dengan mengetahui hal tersebut banyak pilihan yang bisa diperoleh untuk masa depan yang lebih baik. Jenis mutasi tertentu pada Retinitis Pigmentosa saat ini sudah dapat disembuhkan meski baru dilakukan di Negara maju. Dengan mengetahui jenis mutasi gen juga dapat mencegah terjadinya kerusakan photoreceptor lebih lanjut, dikarenakan jenis mutasi gen tertentu dengan pemberian suplemen vitamin A justru memperberat keadaan Retinitis pigmentosanya.

Tindakan yang akan dialami Bapak/ Ibu :

1. Kami akan memberikan pertanyaan mengenai gejala penuruan tajam penglihatan, rabun senja, penyempitan lapang pandang serta seputar anggota keluarga yang menderita keluhan yang sama dengan penderita.

2. Kami akan melakukan pemeriksaan :

    a. Tajam penglihatan dengan menggunakan optotipe Snellen, hitung jari, lambaian tangan dan sentolop.
    b. Lapang pandang dengan menggunakan tes konfrontasi ataupun Humprey visual analysis..
    c. Funduscopy dengan menggunakan indirek oftalmoskop
    d. Pengambilan darah perifer sebanyak 10 cc dengan menggunakan spuit dan dimasukkan dalam tabung yang mengandung EDTA

3. Keuntungan yang diperoleh penderita :
    - Penderita mengetahui gen penyebab dan sifat penurunannya daripada Retinitis pigmentosa yang dideritanya sehingga kemungkinan penurunan gen tersebut pada generasi selanjutnya dapat diketahui. Hal ini sangat penting apabila penderita menginginkan keturunan ataupun rencana pernikahan dikemudian hari.
    - Terapi dapat diberikan sesuai dengan jenis mutasinya meski masih terbatas pada

jenis mutasi tertentu .

- Tidak membahayakan penderita

Penyakit Bapak/Ibu akan dirahasiakan dan rahasia akan kami jaga atas seluruh data penelitian ini.

Nama : A Kentar Arimadyo Sulakso
Alamat/ HP : Jl. Bukit Barisan Perum Permata Puri Ngaliyan 50189, Semarang / 08886532130

Kalau Bapak/ Ibu tidak bersedia ikut dalam penelitian ini, Bapak/ Ibu bebas menolak. Atau apabila Bapak/ Ibu menghendaki mengundurkan diri dari penelitian ini, kami akan menghormati keinginan tersebut.

Atas kerjasama dari Bapak/ Ibu kami mengucapkan terimakasih.

Setelah mendengar dan memahami penjelasan penelitian , dengan ini saya menyatakan

SETUJU/ TIDAK SETUJU

Untuk ikut sebagai responder/ sampel penelitian.

Semarang,

Saksi,

(……..) (……..)

Alamat : Alamat :
Telp : Telp :
Tanggal Pemeriksaan : No. Pasien :

Appendix 5

FORMAT STATUS PENELITIAN
PEMERIKSAAN AWAL

1. No. Register : Alamat Rumah :
2. Nama : No. Telp :
3. Jenis Kelamin :
4. Umur :
5. Pendidikan :
6. Pekerjaan :
7. Kapan mulai timbulnya penurunan tajam penglihatan :
8. Kapan mulai timbulnya rabun senja :
9. Kapan penyakit yang diderita di diagnosis sebagai RP :
10 Apakah ada saudara lain yang menderita penyakit seperti ini? Sebutkan
11 Apakah ada pernikahan antar saudara dekat?

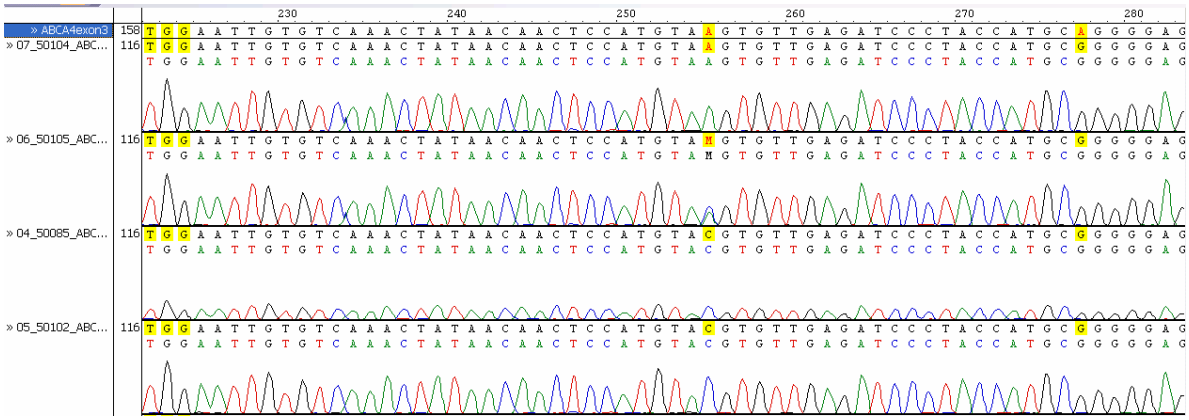
Pemeriksaan :

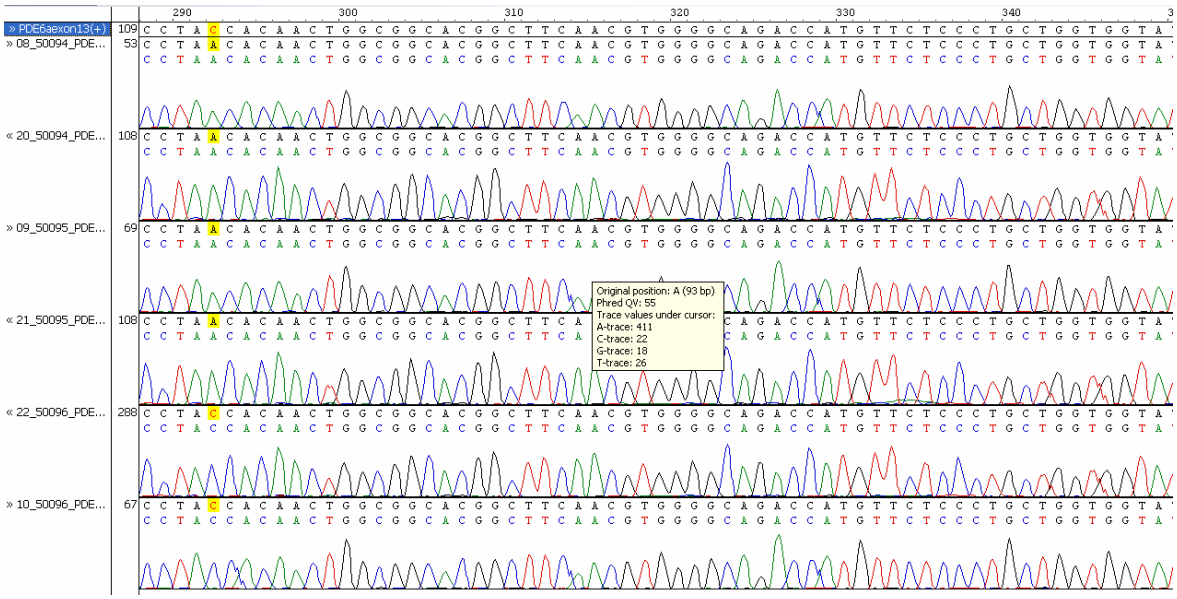
	Mata Kanan	**Mata Kiri**
Visus		

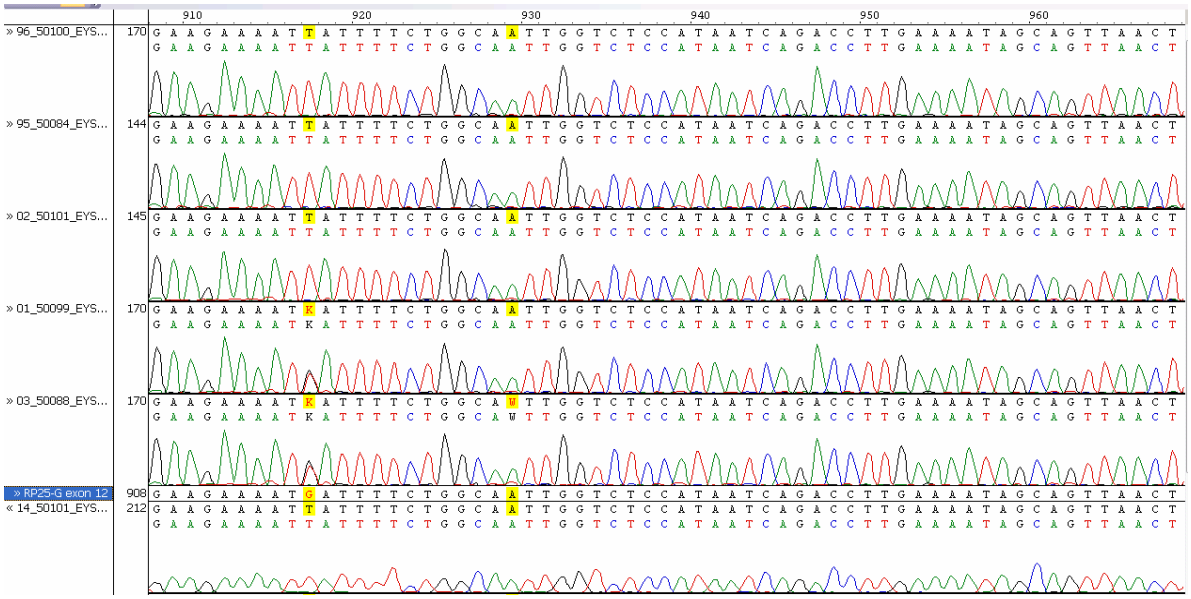
Segmen Anterior
Funduscopy

Appendix 6

Segregation among family




290
300
310
320
330
340
» PDE6aexon13(+)
» 08_50094_PDE...
« 20_50094_PDE...
» 09_50095_PDE...
« 21_50095_PDE...
« 22_50096_PDE...
» 10_50096_PDE...
Original position: A (93 bp)
Phred QV: 55
Trace values under cursor:
A-trace: 411
C-trace: 22
G-trace: 18
T-trace: 26


910
920
930
940
950
960
» 96_50100_EYS...
» 95_50084_EYS...
» 02_50101_EYS...
» 01_50099_EYS...
» 03_50088_EYS...
» RP25-G exon 12
« 14_50101_EYS...

Appendix 7

Laboratory Method

DNA Extraction

DNA was isolated with a salt saturation method as follow:

EDTA frozen blood was transferred into a 50 mL tube. NH4CL 5-10 ml lysis buffer was added to the tube and incubated for 10 - 30 minutes at room temperature. Then the tube was centrifuged for 5 minutes at 3000 -3500 RPM, the supernatant was removed and NH4CL lysis buffer was added again. These steps were repeated three times. Two milliliters of TE lysis buffer, Proteinase-K 10 mg/mL and 100 ul 10% SDS were added and mixed gently into a white pellet and then incubated at 50 degree Celsius for 24 hours. Subsequently NaCl 6M approximately one third volume of the tube was added to the suspension and centrifuged at 4000 RPM for 10 minutes. New tubes were used for the supernatant and absolute ethanol twice volume supernatant was added. DNA that looked like white substance was removed by fine needle. After that, DNA was rinsed with 70% ethanol and transferred into a 1,5 ml tube. Excess ethanol was evaporated by leaving the tube open for at least 1 hour. Then the DNA was dissolved into TE buffer