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LAMPIRAN

1. Persetujuan Komite Etik

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RSPTN UNIVERSITAS HASANUDDIN
RSUP Dr. WAHIDIN SUDIROHUSODO MAKASSAR
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REKOMENDASI PERSETUJUAN ETIK
Nomor : 267/UN4.6.4.5.31/ PP36/ 2020

Tanggal: 11 Mei 2020

Dengan ini Menyatakan bahwa Protokol dan Dokumen yang Berhubungan Dengan Protokol berikut ini telah mendapatkan Persetujuan Etik :

No Protokol	UH20020135	No Sponsor Protokol	
Peneliti Utama	**dr. Marlyanti Nur Rahmah Akib,Sp.M(K), M.Kes**	Sponsor	
Judul Peneliti	Mutasi dan Ekspresi mRNA Gen PITX-3 dan CRYAA pada Penderita Katarak Kongenital di Sulawesi Selatan		
No Versi Protokol	**2**	Tanggal Versi	**4 Mei 2020**
No Versi PSP	**2**	Tanggal Versi	**4 Mei 2020**
Tempat Penelitian	**RS UNHAS, RS dr. Wahidin Sudirohusodo, Klinik Mata Orbita Makassar**		
Jenis Review	[ ] Exempted [x] Expedited [ ] Fullboard Tanggal	Masa Berlaku **11 Mei 2020** sampai **11 Mei 2021**	Frekuensi review lanjutan
Ketua Komisi Etik Penelitian Kesehatan FKUH	Nama **Prof.Dr.dr. Suryani As'ad, M.Sc.,Sp.GK (K)**	Tanda tangan	
Sekretaris Komisi Etik Penelitian Kesehatan FKUH	Nama **dr. Agussalim Bukhari, M.Med.,Ph.D.,Sp.GK (K)**	Tanda tangan	

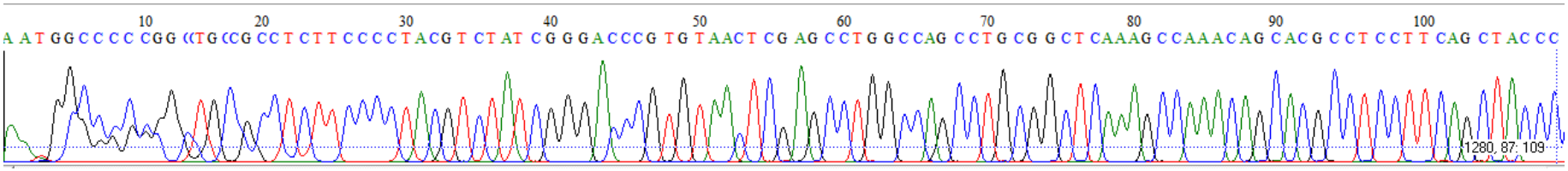
Kewajiban Peneliti Utama:
- Menyerahkan Amandemen Protokol untuk persetujuan sebelum di implementasikan
- Menyerahkan Laporan SAE ke Komisi Etik dalam 24 Jam dan dilengkapi dalam 7 hari dan Lapor SUSAR dalam 72 Jam setelah Peneliti Utama menerima laporan
- Menyerahkan Laporan Kemajuan (progress report) setiap 6 bulan untuk penelitian resiko tinggi dan setiap setahun untuk penelitian resiko rendah
- Menyerahkan laporan akhir setelah Penelitian berakhir
- Melaporkan penyimpangan dari prokol yang disetujui (protocol deviation / violation)
- Mematuhi semua peraturan yang ditentukan

2. Master Tabel Pasien

NO	Nama	Tgl. Lahir/ Umur (thn)	Jenis Katarak	Unilateral/ Bilateral	Mikrof-talmia	Kalsifikasi Kapsul anterior	Fibrosis Kapsul posterior	Ig G Ant.Toxo (<8 IU/ml)	Ig M Ant.Toxo (<0,65 COI)	Ig G Ant.Rubella (<14 IU/ml)	Ig M Ant.Rubella (<1,2 COI)	Ig G Ant.CMV (<6 IU/ml)	Ig M Ant.CMV (<0,9 COI)	Gen PITX3				
														Ekspresi mRNA gen PITX3				Mutasi gen PITX3
														Darah	Kapsul Lensa	Massa Lensa	Humor Aquous	
1	Alfatih	2	Nuklear	Bilateral	(-)	(-)	(-)	NR	NR	>192	NR	NR	>36	0.003948	0.00725	0.000439	0.000568	Normal
2	A. Luthfia	6	Total	Bilateral	(+)	(-)	(-)	>300	NR	>132	NR	NR	NR	0.000524	0.130289	0.000935	0.008339	Normal
3	Yumna	4	Membranous	Unilateral	(+)	(-)	(+)	NR	NR	>93	NR	NR	>62	0.001321	0.001229	0.000951	0.002734	Mutasi/Delesi
4	M. Jafar	11	Nuklear	Bilateral	(-)	(-)	(-)	NR	NR	317	NR	41	NR	0.170731	0.006552	0.000152	0.013754	Mutasi/Delesi
5	Esterlien Deva	6	Nuklear	Bilateral	(-)	(-)	(-)	NR	NR	> 400	NR	42	NR	0.020653	0.000985	0.00157	0.002469	Normal
6	Aditia	7	Membranous	Unilateral	(-)	(+)	(+)	NR	NR	389	NR	30	NR	8.184311	0.055938	0.000393	0.008187	Normal
7	Silvia	6	Nuklear	Bilateral	(-)	(+)	(+)	NR	NR	NR	0.1	0.54	0.07	0.361707	0.001588	0.001581	0.002762	Normal
8	Nirwana	8	Nuklear	Bilateral	(-)	(-)	(-)	>300	NR	NR	NR	74	NR	21.05319	0.000842	0.000305	0.569773	Normal
9	Musdalifah	4	Nuklear	Bilateral	(-)	(-)	(+)	NR	NR	223	NR	42	NR	0.481366	0.00097	0.005141	0.044952	Mutasi/Delesi
10	Afiqa	3	Nuklear	Bilateral	(-)	(-)	(+)	NR	NR	NR	NR	68	NR	35.00103	0.003485	0.001184	0.014162	Normal
11	Arsya	1	Membranous	Unilateral	(-)	(-)	(+)	NR	NR	151	NR	NR	NR	0.026217	0.002792	0.012908	0.20153	Normal
12	Muh. Zahrul	1	Total	Bilateral	(-)	(-)	(-)	NR	NR	101	NR	59	NR	0.132288	0.008843	0.028318	0.000657	Normal
13	Aril	1	Total	Bilateral	(-)	(-)	(-)	NR	NR	199	NR	42	NR	0.020122	0.001998	0.026004	0.019925	Normal
14	Gilang Mandala	1	Total	Unilateral	(-)	(+)	(-)	NR	NR	NR	NR	NR	NR	0.002281	0.008585	0.004925	0.02424	Normal
15	Waode Karunia	5	Nuklear	Bilateral	(+)	(-)	(+)	NR	NR	128	NR	42	NR	0.012773	0.001567	0.002913	0.000382	Normal

3. Hasil Elektroforegram

Kontrol



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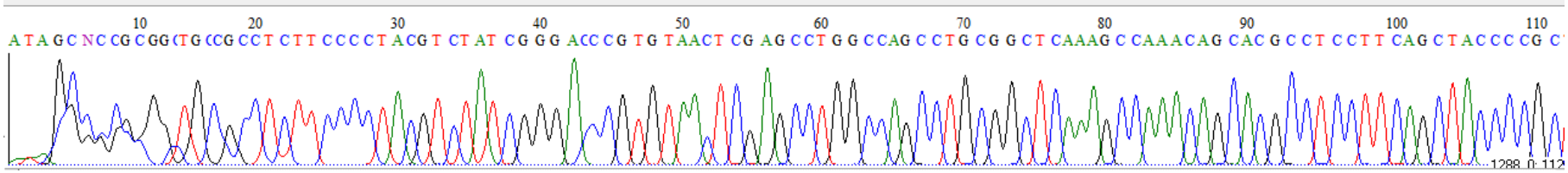

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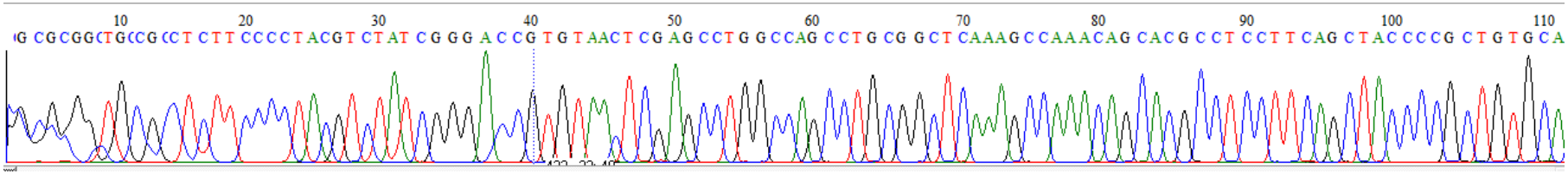

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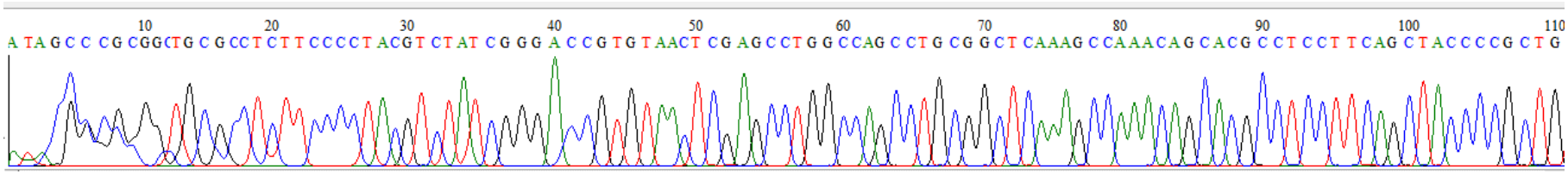

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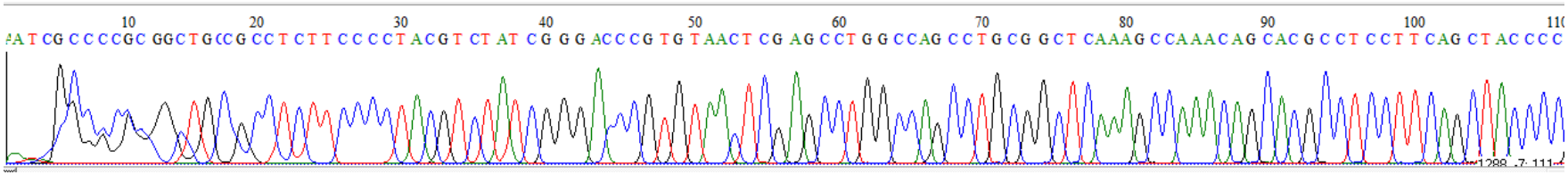

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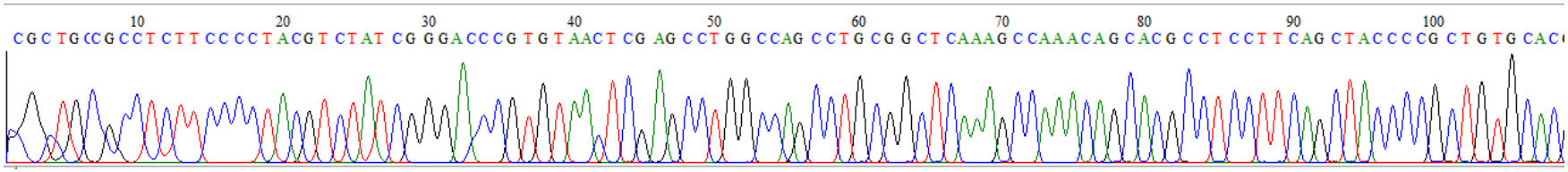

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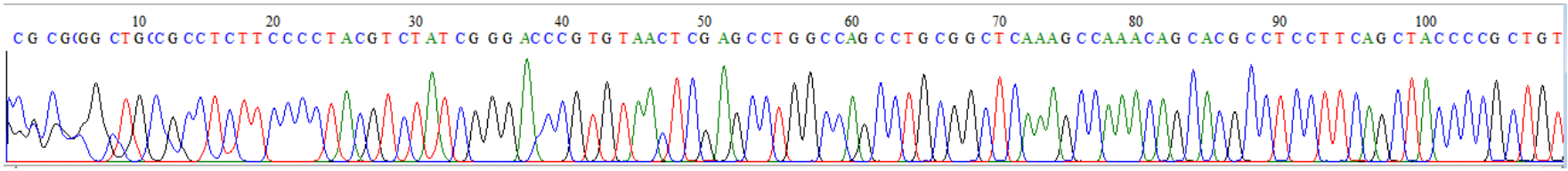

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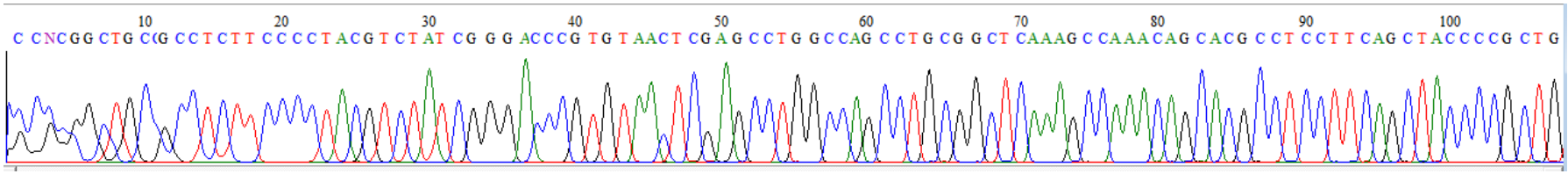

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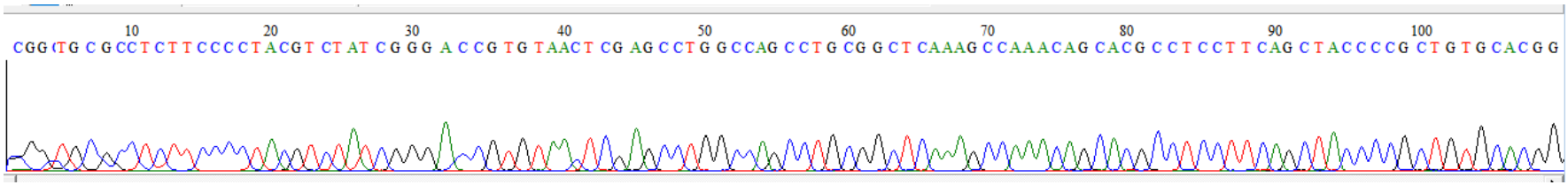

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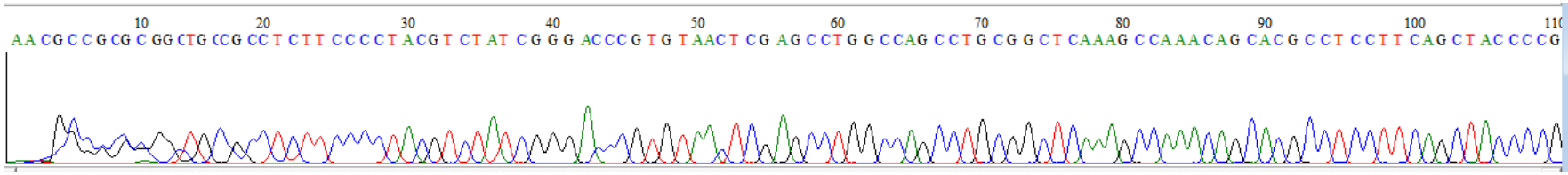

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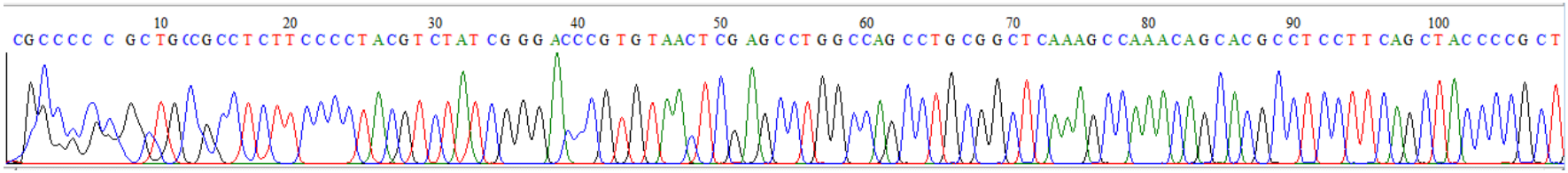

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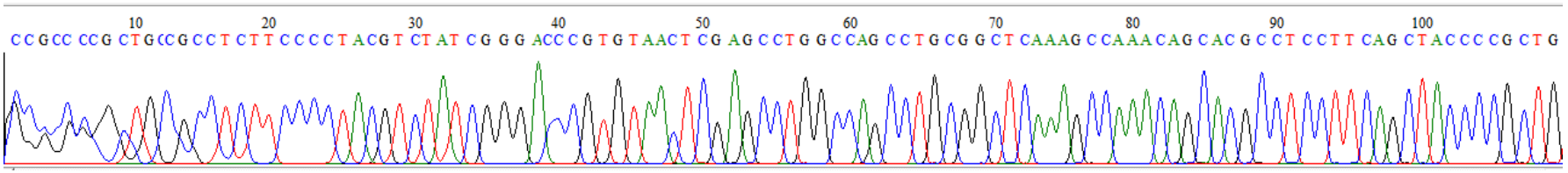

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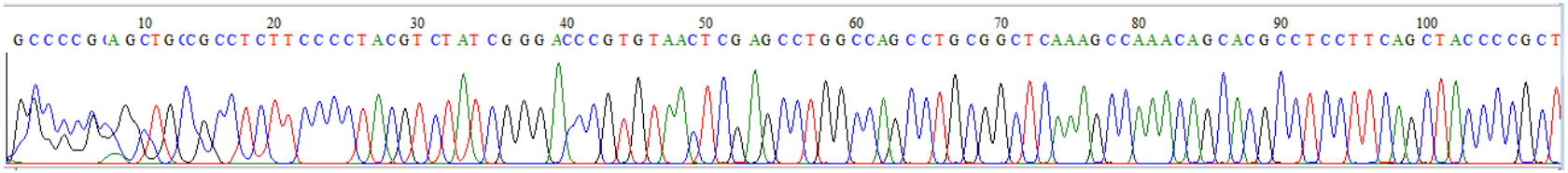

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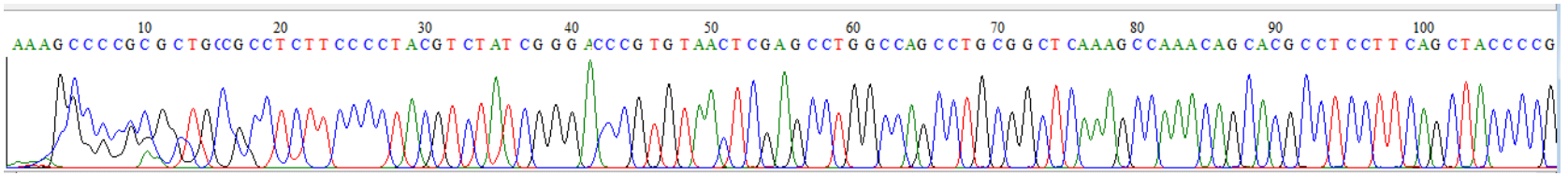

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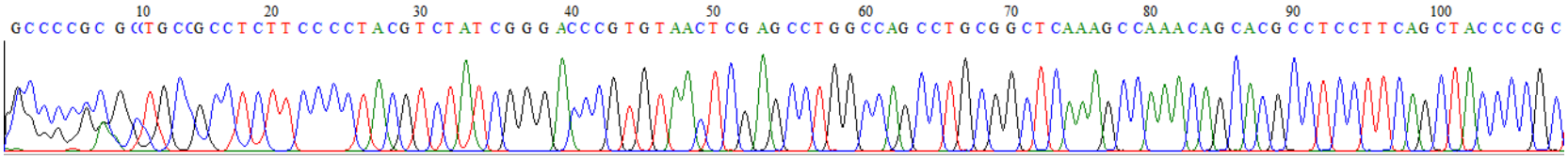

Homo sapiens paired like homeodomain 3 (PITX3), mRNA

NCBI Reference Sequence: NM_005029.4

FASTA Graphics

Go to:

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DEFINITION  Homo sapiens paired like homeodomain 3 (PITX3), mRNA.
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VERSION     NM_005029.4
KEYWORDS    RefSeq; MANE Select.
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
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            Catarrhini; Hominidae; Homo.
REFERENCE   1  (bases 1 to 1426)
  AUTHORS   Wu Z, Meng D, Fang C, Li J, Zheng X, Lin J, Zeng H, Lv S, Zhang Z,
            Luan B, Zhong Z and Chen J.
  TITLE     PITX3 mutations associated with autosomal dominant congenital
            cataract in the Chinese population
  JOURNAL   Mol Med Rep 19 (4), 3123-3131 (2019)
   PUBMED   30816539
  REMARK    GeneRIF: The functional analysis of these 2 PITX3 mutations in the
            in vitro functional studies is an important complement and
            extension, which provides a potential interpretation for the
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pathogenesis and molecular mechanism of PITX3 mutations associated with CC.

REFERENCE 2 (bases 1 to 1426)
AUTHORS Fan Q, Li D, Cai L, Qiu X, Zhao Z, Wu J, Yang J and Lu Y.
TITLE A novel mutation in the OAR domain of PITX3 associated with congenital posterior subcapsular cataract
JOURNAL BMC Med. Genet. 20 (1), 42 (2019)
PUBMED 30894134
REMARK GeneRIF: The mutation c.797_814del, p.Ser266_Ala271del is a novel mutation in the conserved DNA-binding OAR domain of PITX3 that causes congenital cataract.
Publication Status: Online-Only

REFERENCE 3 (bases 1 to 1426)
AUTHORS Zazo Seco C, Plaisancie J, Lupasco T, Michot C, Pechmeja J, Delanne J, Cottereau E, Ayuso C, Corton M, Calvas P, Ragge N and Chassaing N.
TITLE Identification of PITX3 mutations in individuals with various ocular developmental defects
JOURNAL Ophthalmic Genet. 39 (3), 314-320 (2018)
PUBMED 29405783
REMARK GeneRIF: Heterozygous mutation in the PITX3 gene is associated with ocular developmental defects.

REFERENCE 4 (bases 1 to 1426)
AUTHORS Backstrom D, Domellof ME, Granasen G, Linder J, Mayans S, Elgh E, Mo SJ and Forsgren L.

TITLE PITX3 genotype and risk of dementia in Parkinson's disease: A population-based study
JOURNAL J. Neurol. Sci. 381, 278-284 (2017)
PUBMED 28991698
REMARK GeneRIF: Results show that a common polymorphism in the PITX3 gene affects the risk of developing Parkinson's disease (PD) dementia and visuospatial dysfunction in idiopathic PD. If validated, these findings can provide new insights into the neurobiology and genetics of non-motor symptoms in PD.
REFERENCE 5 (bases 1 to 1426)
AUTHORS Yin Y, Morgunova E, Jolma A, Kaasinen E, Sahu B, Khund-Sayeed S, Das PK, Kivioja T, Dave K, Zhong F, Nitta KR, Taipale M, Popov A, Ginno PA, Domcke S, Yan J, Schubeler D, Vinson C and Taipale J.
TITLE Impact of cytosine methylation on DNA binding specificities of human transcription factors
JOURNAL Science 356 (6337) (2017)
PUBMED 28473536
REFERENCE 6 (bases 1 to 1426)
AUTHORS Berry V, Yang Z, Addison PK, Francis PJ, Ionides A, Karan G, Jiang L, Lin W, Hu J, Yang R, Moore A, Zhang K and Bhattacharya SS.
TITLE Recurrent 17 bp duplication in PITX3 is primarily associated with posterior polar cataract (CPP4)
JOURNAL J. Med. Genet. 41 (8), e109 (2004)
PUBMED 15286169
REMARK GeneRIF: A family with posterior polar cataract with a novel

deletion mutation in PITX3.
REFERENCE 7 (bases 1 to 1426)
AUTHORS Nelson KA and Witte JS.
TITLE Androgen receptor CAG repeats and prostate cancer
JOURNAL Am. J. Epidemiol. 155 (10), 883-890 (2002)
PUBMED 11994226
REMARK GeneRIF: Meta-analysis and HuGE review of genotype prevalence and gene-disease association. (HuGE Navigator)
Review article
REFERENCE 8 (bases 1 to 1426)
AUTHORS Semina EV, Ferrell RE, Mintz-Hittner HA, Bitoun P, Alward WL, Reiter RS, Funkhauser C, Daack-Hirsch S and Murray JC.
TITLE A novel homeobox gene PITX3 is mutated in families with autosomal-dominant cataracts and ASMD
JOURNAL Nat. Genet. 19 (2), 167-170 (1998)
PUBMED 9620774
REFERENCE 9 (bases 1 to 1426)
AUTHORS Bardakjian,T., Weiss,A. and Schneider,A.
TITLE Microphthalmia/Anophthalmia/Coloboma Spectrum - ARCHIVED CHAPTER, FOR HISTORICAL REFERENCE ONLY
JOURNAL (in) Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K and Amemiya A (Eds.);
GENEREVIEWS((R));
(1993)
PUBMED 20301552

REFERENCE 10 (bases 1 to 1426)
AUTHORS Hittner,H.M., Kretzer,F.L., Antoszyk,J.H., Ferrell,R.E. and Mehta,R.S.
TITLE Variable expressivity of autosomal dominant anterior segment mesenchymal dysgenesis in six generations
JOURNAL Am. J. Ophthalmol. 93 (1), 57-70 (1982)
PUBMED 6801987
COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff. The reference sequence was derived from AL160011.35.
This sequence is a reference standard in the RefSeqGene project.
On Nov 26, 2018 this sequence version replaced NM_005029.3.

Summary: This gene encodes a member of the RIEG/PITX homeobox family, which is in the bicoid class of homeodomain proteins. Members of this family act as transcription factors. This protein is involved in lens formation during eye development. Mutations of this gene have been associated with anterior segment mesenchymal dysgenesis and congenital cataracts. [provided by RefSeq, Jul 2008].

Publication Note: This RefSeq record includes a subset of the publications that are available for this gene. Please see the Gene record to access additional publications.

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            RNAseq introns              :: mixed/partial sample support
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                                           [ECO:0000350]
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            RefSeq Select criteria :: based on single protein-coding transcript
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            COMPLETENESS: full length.
PRIMARY     REFSEQ_SPAN         PRIMARY_IDENTIFIER PRIMARY_SPAN        COMP
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            181-310             AL160011.35        17286-17415         c
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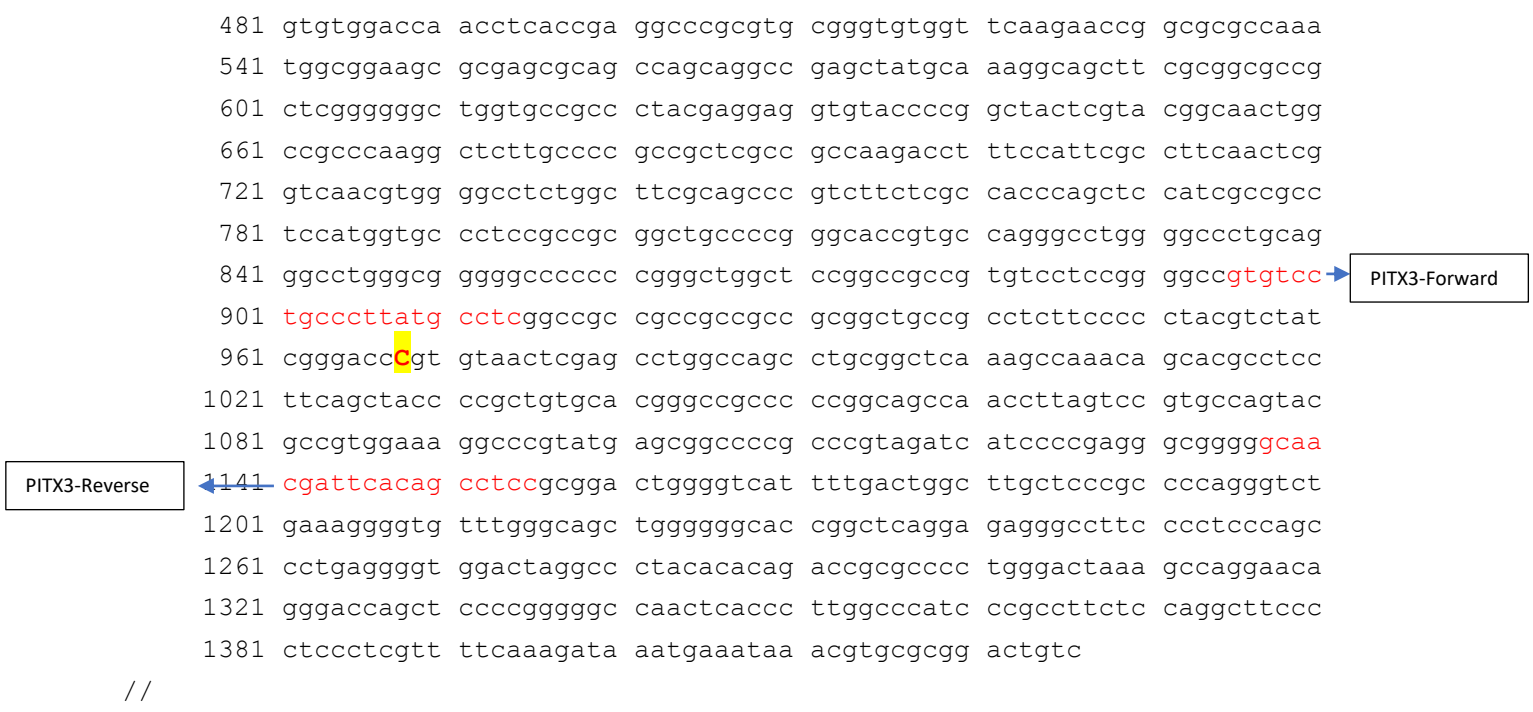
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//
PITX3-Forward
PITX3-Reverse

4. Daftar Riwayat Hidup

A. Data Pribadi

Nama	: Marlyanti Nur Rahmah
NIDN	: 0916108001
Tempat & Tanggal Lahir	: Palu, 16 Oktober 1980
Agama	: Islam
Orang Tua	: dr. Muhammad Akib Kamaluddin
	dr. Mariani Akib Baramuli, MM
Suami	: dr. Tommy Rubiyanto, Sp.B, Sp.BA
Anak	: Muhammad Daffa Tsaqif Naufal
	Dafinah Shafa Malikah
Pekerjaan	: Dosen
Alamat	: Citraland Celebes, Cluster Maroon Vogue Blok F9/26

B. Riwayat Pendidikan

No	Jenis/ Jurusan Pendidikan	Nama Sekolah/ Institusi	Tahun Lulus
1	SD	SD Negeri Inti Palu	1992
2	SMP	SMP Negeri 1 Palu	1995
3	SMA	SMA Negeri 2 Palu	1998
4	S1	Fakultas Kedokteran UMI	2002
5	Profesi Dokter	Fakultas Kedokteran UMI	2004
6	S2	PPDS Ilmu Kesehatan Mata	2010
7	S3	Ilmu Kedokteran Unhas	

C. Riwayat Pekerjaan

1. Dosen Tetap Fakultas Kedokteran UMI tahun 2006 – sekarang
2. Dosen Luar Biasa Departemen Ilmu Kesehatan Mata tahun 2015-sekarang

D. Pelatihan

1. Fellowship Pediatric Ophthalmology dan Strabismus di RS Mata Cicendo
2. Fellowship Pediatric Ophthalmology and Strabismus di Aravind Eye Hospital, Madurai, India
3. Fellowship Pediatric Ophthalmology and Strabismus di Ispahani Islamiah Eye Institute and Hospital, Dhaka, Bangladesh

E. Publikasi Ilmiah

1. Ocular Proptosis as an initial presentation of neuroblastoma in infant : a rare case report dalam Journal of Case Reports and Images in Ophthalmology, Volume 1, 2018
2. Inversting to improve conditions for retention and satisfaction at a paediatric eye centre in South Sulawesi, Indonesia dalam Community Eye Health Journal, Volume 31, 2018
3. mRNA expression of PITX3 gene in congenital cataract caused by rubella virus infection dalam Indian Journal of Forensic Medicine & Toxicology, Volume 14, 2020