

Manuscript Title: The role of genetics in preterm birth

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Online Resource 1: Candidate genes grouped by biological pathway

The table is grouped based on biological pathway and ordered alphabetically based on gene symbol. The table includes references of case-control or cohort candidate gene studies (reference list at end of document) that identified significant polymorphisms within the listed genes that were associated with PTB using maternal samples only, infant samples only, or both maternal and infant samples. The list of genes was extracted from Online Resource 2 (ESM_2.xlsx). Genes are listed as gene symbol and grouped by pathway based on summary of gene function as per (Stelzer et al. 2016). Genes that could not be grouped into one of the first four pathways were allocated into the “Miscellaneous” group.

Pathway	Gene	Maternal only	Infant only	Both
Immunological and Inflammatory	<i>ALOX5</i>	(Liu et al. 2012)		
	<i>ALOX5AP</i>	(Liu et al. 2012)		
	<i>CARD15</i>	(Ryckman et al. 2010)		
	<i>CCL2</i>	(Velez et al. 2008a)		
	<i>CCR2</i>		(Romero et al. 2010a)	
	<i>CD55</i>		(Romero et al. 2010a)	
	<i>C4BPA</i>	(McElroy et al. 2013)		
	<i>CR1</i>	(McElroy et al. 2013)		
	<i>CSF1</i>	(Romero et al. 2010a)		
	<i>CTLA4</i>	(Velez et al. 2008a)		
	<i>DEFB1</i>	(Romero et al. 2010a)		
	<i>FAS</i>	(Fuks et al. 2005)		
	<i>FcγRIIb</i>	(Iwanaga et al. 2011)		
	<i>HLA-DQA1</i>	(Falah et al. 2013)		
	<i>HLA-DRA</i>	(Falah et al. 2013)		
	<i>HLA-DRB1</i>	(Falah et al. 2013)		
	<i>ICAM-1</i>	(Kwon et al. 2009)		
	<i>IFNγ</i>			(Speer et al. 2006)
	<i>IFNGR1</i>		(Romero et al. 2010b)	
	<i>IFNGR2</i>	(Harmon et al. 2013)		
	<i>IL1α</i>	(Sata et al. 2008)	(Velez et al. 2008a)	
	<i>IL1β</i>	(Hollegaard et al. 2008; Belousova et al. 2019; Pereyra et al. 2016)	(Yılmaz et al. 2012) (Velez et al. 2008a; Genç et al. 2002)	
	<i>IL1R1</i>	(Velez et al. 2008a)		

	<i>IL1R2</i>	(Langmia et al. 2015b) (Velez et al. 2008a)		
	<i>IL1RAP</i>			(Velez et al. 2008a)
	<i>IL1RN</i>	(Gillespie et al. 2017) (Velez et al. 2008a)		
	<i>IL2</i>		(Romero et al. 2010b)	
	<i>IL2RA</i>	(Velez et al. 2008a)		
	<i>IL2RB</i>		(Velez et al. 2008a; Velez et al. 2009)	
	<i>IL4</i>	(Harmon et al. 2013; Annells et al. 2004)	(Velez et al. 2008a)	
	<i>IL4R</i>			(Velez et al. 2008a)
	<i>IL5</i>			(Velez et al. 2008a)
	<i>IL6</i>	(Karakas et al. 2018; Gómez et al. 2010; Pereyra et al. 2016; Simhan et al. 2003)	(Fortunato et al. 2008)	
	<i>IL6R</i>	(Velez et al. 2008b; Velez et al. 2007; Fortunato et al. 2008; Velez et al. 2008a)	(Romero et al. 2010b)	
	<i>IL8RA</i>	(Ryckman et al. 2010)		
	<i>IL10</i>	(Han et al. 2020; Lyubomirskaya et al. 2020a; Lyubomirskaya et al. 2020b; Pandey et al. 2018; Annells et al. 2004; Velez et al. 2008a)		(Ramos et al. 2016)
	<i>IL10RA</i>			(Velez et al. 2008a)
	<i>IL10RB</i>		(Ryckman et al. 2010)	
	<i>IL12α</i>	(Harmon et al. 2013)		
	<i>IL13</i>	(Harmon et al. 2013)	(Heinzmann et al. 2009)	
	<i>IL15</i>	(Velez et al. 2009)		
	<i>IL16</i>	(Frey et al. 2016)		
	<i>IL18</i>	(Velez et al. 2008a)	(Krueger et al. 2011)	
	<i>IL18BP</i>		(Romero et al. 2010a)	
	<i>IL23R</i>	(Falah et al. 2013)		
	<i>KIR3DL2</i>	(Harmon et al. 2013)		
	<i>LTA</i>	(Engel et al. 2005)		
	<i>LTF</i>	(Romero et al. 2010b)		
	<i>MBL2</i>	(Silva et al. 2020; Annells et al. 2004)	(Bodamer et al. 2006)	
	<i>MCP1</i>		(Wang et al. 2015)	
	<i>MICA</i>		(Song et al. 2017)	
	<i>NFKB1</i>			(Ryckman et al. 2010)
	<i>NFKBIA</i>			(Velez et al. 2008a)
	<i>NFKBIB</i>			(Velez et al. 2008a)
	<i>NFKBIE</i>	(Ryckman et al. 2010)		
	<i>NOS2A</i>		(Gibson et al. 2007)	
	<i>PLAT</i>			(Velez et al. 2008a)
	<i>PLA2G4A</i>	(Velez et al. 2008a)		
	<i>PTGES</i>	(Liu et al. 2012)		
	<i>PTGES2</i>	(Liu et al. 2012)		
	<i>SFTPD</i>		(Karjalainen et al. 2012a)	
	<i>TIRAP</i>	(Karody et al. 2013)		

	<i>TLR1</i>	(Romero et al. 2010a)	(Romero et al. 2010b)	
	<i>TLR2</i>	(Ramos et al. 2016; Velez et al. 2008a; Romero et al. 2010b)		
	<i>TLR4</i>		(Bitner et al. 2013; Lorenz et al. 2002)	
	<i>TLR7</i>	(Velez et al. 2008a)		
	<i>TNFα</i>	(As Sayaril et al. 2018; Drews-Piasecka et al. 2014; Gebhardt et al. 2009; Harmon et al. 2013; Harper et al. 2011; Hollegaard et al. 2008; Amory et al. 2004; Macones et al. 2004; Roberts et al. 1999)	(Aidoo et al. 2001; Chen et al. 2003; Fortunato et al. 2008; Menon et al. 2006b)	(Yilmaz et al. 2012)
	<i>TNFR1</i>	(Menon et al. 2006b; Menon et al. 2006a)		(Fortunato et al. 2008)
	<i>TNFR2</i>	(Jones et al. 2012; Fortunato et al. 2008; Menon et al. 2006b; Menon et al. 2006a)		
	<i>TREM1</i>		(Velez et al. 2008a)	
Tissue remodelling	<i>AR</i>		(Karjalainen et al. 2012b)	
	<i>B2AR</i>	(Doh et al. 2004)		
	<i>CGA</i>	(Plunkett et al. 2011)		
	<i>COL1A1</i>	(Velez et al. 2008a)		
	<i>COL1A2</i>	(Ryckman et al. 2010)		(Velez et al. 2008a)
	<i>COL3A1</i>			(Velez et al. 2008a)
	<i>COL4A1</i>		(Romero et al. 2010b)	
	<i>COL4A2</i>			(Romero et al. 2010b)
	<i>COL4A3</i>		(Gimenez et al. 2017)	
	<i>COL4A4</i>			(Romero et al. 2010b)
	<i>COL4A5</i>		(Romero et al. 2010b)	
	<i>COL5A1</i>	(Velez et al. 2008a)		
	<i>COL5A2</i>			(Velez et al. 2008a)
	<i>CRH</i>		(Ryckman et al. 2010)	
	<i>CRHBP</i>			(Velez et al. 2008a)
	<i>CRHR2</i>	(Schmid et al. 2010)	(Velez et al. 2008a)	
	<i>CSPG2</i>	(Romero et al. 2010a)		
	<i>FGF1</i>		(Preda et al. 2020)	(Romero et al. 2010b)
	<i>FGF4</i>	(Romero et al. 2010a)		
	<i>FN1</i>		(Romero et al. 2010b)	
	<i>FSHR</i>	(Chun et al. 2013; Plunkett et al. 2011)	(Dominguez-Lopez et al. 2018)	
	<i>IGF2</i>		(Romero et al. 2010b)	
	<i>MMP1</i>		(Wang et al. 2008)	(Velez et al. 2008a)
	<i>MMP3</i>			(Velez et al. 2008a)
	<i>MMP8</i>		(Velez et al. 2008a)	
	<i>MMP9</i>	(Jones et al. 2012; Pandey and Awasthi 2020)		
	<i>MMP10</i>	(Romero et al. 2010a; Romero et al. 2010b)		
	<i>MMP16</i>	(Romero et al. 2010b)		
	<i>MMP19</i>		(Romero et al. 2010a)	

	<i>PGR</i>	(Langmia et al. 2015c; Manuck et al. 2010)	(Velez et al. 2008a)	(Kadivnik et al. 2022)
	<i>PGRMC1</i>	(Velez et al. 2008a)		
	<i>RLN2</i>	(Lyubomirskaya et al. 2020a; Lyubomirskaya et al. 2020b; Rocha et al. 2013; Vogel et al. 2009)		
	<i>SERPINH1</i>		(Wang et al. 2006)	
	<i>TIMP1</i>		(Romero et al. 2010b)	
	<i>TIMP2</i>	(Romero et al. 2010a; Frey et al. 2016)		(Romero et al. 2010b)
	<i>TIMP3</i>			(Velez et al. 2008a)
	<i>TIMP4</i>		(Ryckman et al. 2010)	
	<i>TNR</i>	(Romero et al. 2010a)	(Romero et al. 2010b)	
Metabolic	<i>ADH1B</i>			(Ryckman et al. 2010)
	<i>CETP</i>		(Romero et al. 2010b)	
	<i>CRHR1</i>		(Gimenez et al. 2017)	
	<i>CYP19A1</i>			(Velez et al. 2008a)
	<i>CYP2D6</i>			(Velez et al. 2008a)
	<i>DHFR</i>	(Velez et al. 2008a)		
	<i>EPHX1</i>	(Ryckman et al. 2010)		
	<i>FADS2</i>	(Liu et al. 2012)		
	<i>GC</i>	(Wang et al. 2021)		
	<i>GSTP1</i>		(Velez et al. 2008a)	
	<i>HSD11B1</i>	(Velez et al. 2008a)		
	<i>HSD17B7</i>		(Ryckman et al. 2010)	
	<i>IGF1</i>	(He et al. 2017)	(Velez et al. 2008a)	
	<i>IGF1R</i>		(Haataja et al. 2011)	
	<i>IRS1</i>		(Romero et al. 2010b)	
	<i>LEP</i>			(Salem et al. 2016)
	<i>LPL</i>	(Falah et al. 2013)		
	<i>LRP2</i>	(Wang et al. 2021)		
	<i>MTHFR</i>	(Hwang et al. 2017; Nan and Li 2015; Tiwari et al. 2015; Valdez et al. 2004)	(Zhu et al. 2015)	
	<i>MTRR</i>	(Engel et al. 2006)		
	<i>NAT1</i>	(Velez et al. 2008a)		
	<i>PLA2G4D</i>	(Liu et al. 2017)		
	<i>PON1</i>		(Gimenez et al. 2017; Ryckman et al. 2010; Velez et al. 2008a)	
	<i>PPARG</i>	(Meirhaeghe et al. 2007)		
	<i>SHMT1</i>	(Engel et al. 2006)		
	<i>SLC23A1</i>		(Velez et al. 2008a)	
	<i>SLC23A2</i>	(Erichsen et al. 2006)		
	<i>TCN2</i>	(Kwon et al. 2021)		
	<i>TSHR</i>			(Velez et al. 2008a)
	<i>VDR</i>	(Barchitta et al. 2018; Javorski et al. 2018; Rosenfeld et al. 2017; Manzon et al. 2014)		(Dutra et al. 2020)
Haematological , vascular, and endothelial	<i>ACE</i>	(Romero et al. 2010a)		
	<i>AGT</i>	(Romero et al. 2010b)		
	<i>AGTR</i>			(Romero et al. 2010b)
	<i>ANG</i>	(Romero et al. 2010a)		
	<i>EDN2</i>		(Velez et al. 2008a)	

	<i>ERG</i>	(Plunkett et al. 2011)		
	<i>F2</i>		(Chen et al. 2007)	
	<i>F2R</i>		(Ryckman et al. 2010)	
	<i>F3</i>		(Gimenez et al. 2017)	
	<i>F5</i>	(Yu et al. 2009; McElroy et al. 2013; Velez et al. 2008a)		
	<i>F7</i>	(Velez et al. 2008a)		
	<i>F10</i>	(Ryckman et al. 2010)		
	<i>FLT1</i>	(Frey et al. 2016)		
	<i>MHC</i>	(Falah et al. 2013)		
	<i>NOS3</i>	(Silva et al. 2020)	(Velez et al. 2008a)	
	<i>NPPA</i>		(Romero et al. 2010b)	
	<i>NPR1</i>		(Romero et al. 2010b)	
	<i>PAR1</i>	(Grisaru-Granovsky et al. 2007)		
	<i>PLAUR</i>		(Ryckman et al. 2010)	
	<i>PLG</i>	(Ryckman et al. 2010)		
	<i>PTGER1</i>	(Romero et al. 2010a)		
	<i>PTGER3</i>	(Ryckman et al. 2010)		(Velez et al. 2008a)
	<i>PTGFR</i>			(Ryckman et al. 2010)
	<i>PTGS1</i>	(Liu et al. 2012; Velez et al. 2008a)		
	<i>PTGS2</i>	(Liu et al. 2012)	(Velez et al. 2008a)	
	<i>REN</i>	(Romero et al. 2010a)		(Romero et al. 2010b)
	<i>SELE</i>	(Romero et al. 2010b)		
	<i>SERPINC1</i>		(Romero et al. 2010b)	
	<i>SERPINE1</i>		(Chen et al. 2007)	
	<i>TBXAS1</i>	(Romero et al. 2010b)		
	<i>TFPI</i>		(Ryckman et al. 2010)	
	<i>THBD</i>		(Gibson et al. 2007)	
	<i>THPO</i>	(Romero et al. 2010b)		
	<i>VEGFA</i>	(Langmia et al. 2015a; Velez et al. 2008a)		
	<i>VEGFC</i>	(Romero et al. 2010b)		
	<i>VWF</i>	(Romero et al. 2010a)		
Miscellaneous	<i>ADRB2</i>		(Gibson et al. 2007)	
	<i>AKAP10</i>	(Langmia et al. 2015d)		
	<i>AP3M2</i>			(Velez et al. 2008a)
	<i>CBS</i>		(Velez et al. 2008a)	
	<i>CCHCR1</i>	(Falah et al. 2013)		
	<i>CDKN2</i>	(Falah et al. 2013)		
	<i>CENTG2</i>	(Plunkett et al. 2011)		
	<i>COMT</i>		(Thota et al. 2012)	
	<i>CPNE4</i>	(Plunkett et al. 2011)		
	<i>DUSP16</i>	(Plunkett et al. 2011)		
	<i>ELN</i>		(Romero et al. 2010b)	
	<i>EMR1</i>	(Plunkett et al. 2011)		
	<i>EPHX2</i>			(Velez et al. 2008a)
	<i>FHIT</i>	(Plunkett et al. 2011)		
	<i>GABRG3</i>	(Plunkett et al. 2011)		
	<i>GNB3</i>	(Romero et al. 2010b)		
	<i>HSPA14</i>		(Velez et al. 2008a)	
	<i>HSPA1B</i>		(Ryckman et al. 2010)	
	<i>HSPA6</i>		(Velez et al. 2008a)	
	<i>HSPG2</i>	(Romero et al. 2010b)		

	<i>HTR2A</i>	(Romero et al. 2010b)		
	<i>IGF2R</i>		(Romero et al. 2010b)	
	<i>IMP5</i>		(Romero et al. 2010a)	
	<i>KCNN3</i>	(Day et al. 2011)	(Gimenez et al. 2017)	
	<i>KCNN4</i>	(Day et al. 2011)		
	<i>KL</i>			(Velez et al. 2008a)
	<i>LDB2</i>	(Plunkett et al. 2011)		
	<i>LMO1</i>	(Plunkett et al. 2011)		
	<i>LPA</i>		(Romero et al. 2010b)	
	<i>MTHFD1</i>	(Velez et al. 2008a)		
	<i>NPAS3</i>	(Plunkett et al. 2011)		
	<i>NR3C1</i>		(Velez et al. 2008a)	
	<i>NR3C2</i>	(Christiaens et al. 2015)		
	<i>PAFAH1B1</i>		(Velez et al. 2008a)	
	<i>PAX9</i>	(Plunkett et al. 2011)		
	<i>PER3</i>	(Kovac et al. 2019)		
	<i>PGEA1</i>		(Ryckman et al. 2010)	
	<i>PGRMC2</i>	(Ryckman et al. 2010)		
	<i>PLA2G4C</i>	(Plunkett et al. 2010)		
	<i>PON2</i>			(Velez et al. 2008a)
	<i>PRKCA</i>	(Frey et al. 2016; Gómez et al. 2010)		
	<i>PTPRD</i>	(Plunkett et al. 2011)		
	<i>RASD2</i>	(Plunkett et al. 2011)		
	<i>RHOBTB3</i>	(Plunkett et al. 2010)		
	<i>SCNN1A</i>	(Velez et al. 2008a)		
	<i>SLC6A4</i>			(Ryckman et al. 2010)
	<i>SEPS1</i>		(Wang et al. 2013)	
	<i>SHP2</i>		(Shim et al. 2017)	
	<i>SKA2</i>	(Ijabi et al. 2019)		
	<i>SLC35B2</i>		(Velez et al. 2008a)	
	<i>TERT</i>	(Marrs et al. 2018)		
	<i>TEX12</i>	(Velez et al. 2008a)		
	<i>TGFB1</i>	(Speer et al. 2006)		
	<i>THBS4</i>	(Romero et al. 2010b)		
	<i>UGT1A1</i>			(Velez et al. 2008a)
	<i>VRK2</i>	(Plunkett et al. 2011)		
	<i>WWOX</i>	(Plunkett et al. 2011)		

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