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| Table S6: Genes shared by patients I1 and II1 but not by patient II7 | | | | | | | | | | | | |
| Gene | Gene function | | Expression | | | | | | | | | |
| lymphatic | endothelial | | Lymph nodes | | Smooth muscle | Dentritic cells | T cells (CD4+) | T cells (CD8+) | B lymphoblasts |
| ANP32B | Multifunctional protein working as a cell cycle progression factor as well as a cell survival factor. | | +++ | +++ | | +++ | | +++ | +++ | +++ | +++ | +++ |
| Required for the progression from the G1 to the S phase. Anti-apoptotic protein which functions as a caspase-3 | |
| inhibitor. Has no phosphatase 2A (PP2A) inhibitor activity (By similarity). Exhibits histone chaperone | |
| properties, stimulating core histones to assemble into a nucleosome | |
| CDC27 | Cell division defect | | + | + | | + | | + | + | + | + | + |
| Increased S DNA content, | |
| Increased cell death HMECs cell | |
| Increased cell death in breast cell | |
| Increased number of mitotic | |
| Synthetic lethal with Ras | |
| CSRP2BP | Component of the ATAC complex, a complex with histone acetyltransferase activity on histones H3 and H4. | | + | + | | + | | + | + | + | + | + |
| May function as a scaffold for the ATAC complex to promote ATAC complex stability. Has also weak histone | |
| acetyltransferase activity toward histone H4. Required for the normal progression through G1 and G2/M phases of the cell cycle | |
| CTBP2 | Corepressor targeting diverse transcription regulators;  Functions in brown adipose tissue (BAT) differentiation | |  | ++ | | + | | ++ | ++ | + | + | ++ |
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| Table S6: Genes shared by patients I1 and II1 but not by patient II7 | | | | | | | | | | | | |
| Gene | Gene function | | Expression | | | | | | | | | |
| lymphatic | endothelial | | Lymph nodes | | Smooth  muscle | Dentritic cells | T cells (CD4+) | T cells (CD8+) | B lymphoblasts |
| DMD | Anchors the extracellular matrix to the cytoskeleton via F-actin. | |  | + | | + | | + | + | + | + | + |
| Ligand for dystroglycan | |
| Component of the dystrophin-associated glycoprotein complex which accumulates at the neuromuscular junction (NMJ) and at a variety of synapses in the peripheral and central nervous systems and has a structural function in stabilizing the sarcolemma. | |
| DPYSL4 | Also implicated in signaling events and synaptic transmission | |  | + | | + | | + | + | + | + | + |
| Necessary for signaling by class 3 semaphorins and subsequent remodeling of the cytoskeleton. | |
| Plays a role in axon guidance, neuronal growth cone collapse and cell migration | |
| FBP1 | Catalytic activity: D-fructose 1,6-bisphosphate + H(2)O = D-fructose 6-phosphate + phosphate | |  | + | | + | | + | + | + | + | + |
| Enzyme regulation: Subject to complex allosteric regulation. | |
| The enzyme can assume an active R-state, or an inactive T-state | |
| Intermediate conformations may exist. AMP acts as allosteric inhibitor. | |
| AMP binding affects the turnover of bound substrate and not the affinity for substrate. | |
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| Table S6: Genes shared by patients I1 and II1 but not by patient II7 | | | | | | | | | | | | |
| Gene | Gene function | Expression | | | | | | | | | | |
| lymphatic | | endothelial | Lymph nodes | | | Smooth  muscle | Dentritic cells | T cells (CD4+) | T cells (CD8+) | B lymphoblasts |
| FLT4 | Promotes proliferation, survival and migration of endothelial cells, and regulates angiogenic sprouting. | + | | ++ | + | | | + | + | + | + | + |
| Signaling by activated FLT4 leads to enhanced production of VEGFC, and to a lesser degree VEGFA, thereby creating a positive feedback loop that enhances FLT4 signaling. |
| Modulates KDR signaling by forming heterodimers. The secreted isoform 3 may function as a decoy receptor for VEGFC and/or VEGFD and play an important role as a negative regulator of VEGFC-mediated lymphangiogenesis and angiogenesis. |
| Binding of vascular growth factors to isoform 1 or isoform 2 leads to the activation of several signaling cascades; |
| Mediates activation of the MAPK1/ERK2, MAPK3/ERK1 signaling pathway, of MAPK8 and the JUN signaling pathway, and of the AKT1 signaling pathway. |
| FRMD3 | Putative tumor suppressor gene that may be implicated in the origin and progression of lung cancer |  | |  |  | |  | |  |  |  |  |
| GGT1 | Cleaves the gamma-glutamyl bond of extracellular glutathione (gamma-Glu-Cys-Gly), glutathione conjugates, and other gamma-glutamyl compounds.  The metabolism of glutathione releases free glutamate and the dipeptide, cysteinyl-glycine, which is hydrolyzed to cysteine and glycine by dipeptidases. |  | |  |  | |  | |  |  |  |
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| |  |  |  |  |  |  |  |  |  |  | | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | | Table S6: Genes shared by patients I1 and II1 but not by patient II7 | | | | | | | | | | | Gene | Gene function | expression | | | | | | | | | lymphatic | endothelial | Lymph nodes | Smooth  muscle | Dentritic cells | T cells (CD4+) | T cells (CD8+) | B lymphoblasts | | GGT1 | Initiates extracellular glutathione (GSH) breakdown, provides cells with a local cysteine supply and contributes to maintain intracellular GSH level. |  |  |  |  |  |  |  |  | | It is part of the cell antioxidant defense mechanism. Isoform 3 seems to be inactive | | immunoglobulin superfamily member 3,highly expressed in placenta,kidney and lung | | IGSF3 | immunoglobulin superfamily member 3,highly expressed in placenta,kidney and lung | + | + | + | + | + | + | + | + | | MCEE | methylmalonyl-CoA epimerase activity | + | + | + | + | + | + | + | + | | PABPC3 | Binds the poly(A) tail of mRNA. May be involved in cytoplasmic regulatory processes of mRNA metabolism. | ++++ | ++++ | ++++ | ++++ | ++++ | ++++ | ++++ | ++++ | | Binds poly(A) with a slightly lower affinity as compared to PABPC1 | | PCDHA13 | Potential calcium-dependent cell-adhesion protein. |  |  |  |  |  |  |  |  | | May be involved in the establishment and maintenance  of specific neuronal connections in the brain | |  |  |  |  |  |  |  |  |  |  | | | | | | | | | | | | | |