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| --- | --- | --- | --- | --- |
| **Gene** | **Alteration in gene expression in DS** | **Protein** | **Alteration in protein expression in DS** | **Metabolic and functional consequences** |
| CBS | Upregulation (XX fold) | Cystathionine b-synthase | Upregulation (XX fold) | Increased H2S was found to directly inhibit mitochondrial Complex IV, which would be directly responsible for the inhibition of aerobic ATP generation. H2S would also be expected to lead to posttranscriptional modification of multiple proteins in DS, which would be expected to significantly affect their function (unrelated to the expression level of the protein). |
| ATP5PF | Upregulation (XX fold) | ATP synthase peripheral stalk subunit F6 | Downregulation (XX fold)**??** | Lower levels of ATP synthase would be expected to produce an impairment of ATP production, which, in fact, is reflected in the reduced ATP levels and energy charge in DS. |
| ATP5PO | Upregulation (XX fold) | ATP synthase peripheral stalk subunit OSCP | Downregulation (XX fold) **??** |
| PFK | Upregulation (XX fold) | phosphofructokinase | Upregulation (XX fold) **??** | Upregulation of these enzymes would be expected to increase glycolysis, perhaps as a compensatory response to the inhibition of aerobic (mitochondrial) ATP generation. These responses can be viewed as complementary parts of cellular pseudohypoxia in DS. |
| G6PDH | Upregulation (XX fold) | glucose-6-phosphate dehydrogenase | Upregulation (XX fold) **??** |
| OGDH | Upregulation (XX fold) | oxoglutarate dehydrogenase | **??**regulation (XX fold) **??** | Itt kellene megnezni a metabolomic cikket es hatha abbol kiderul, merre megy a citrat kor es mi a hatasa ezeknek az expresszios valtozasoknak |
| ACLY | Upregulation (XX fold) | ATP citrate lyase | **??**regulation (XX fold) **??** |
| **??** |  |  |  |  |
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