Robust phenotyping of developmental defects using convolutional neural networks

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Early development is orchestrated by a handful of signaling pathways. Reducing or abolishing the activity of these signaling pathways leads to characteristic developmental defects. While the classification of phenotypic defects can help to identify the underlying signaling mechanisms, this requires expert knowledge and the classification schemes have not been standardized. We have developed a machine learning approach based on convolutional neural networks for automated phenotyping by training computer vision algorithms to identify signaling mutants. Combined with a model of time-dependent developmental trajectories, this approach identifies and classifies with high precision phenotypic defects caused by loss-of-function of the seven major signaling pathways relevant for vertebrate development. Our classification algorithms can also robustly identify defects caused by different combinations in the dosage of signaling pathway activity. This approach has wide applications in developmental biology and automated phenotyping in high-throughput drug screens.